



Supplementary Materials for

Systematic Localization of Common Disease-Associated Variation in Regulatory DNA

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Materials and Methods

1.1. Disease- and trait-associated variants from GWAS

The GWAS SNP set used for analysis was derived from the NHGRI GWAS Catalog, downloaded on January 4, 2012 (2). The catalog is a continually-updated compendium of GWAS which lists the single SNP from each gene or region with the strongest disease association identified by the studies. Each study attempted to assay at least 100,000 SNPs across the genome. The catalog contained 6,896 entries at the time of download. We excluded SNPs mapping outside the main chromosome contigs, including the "random" chromosome fragments, SNPs without coordinates in the GRCh37/hg19 human genome assembly, SNPs without a dbSNP ID, and records which were a combination of multiple SNPs associated with a disease or trait. The catalog contained data from 920 publications mapping 679 total diseases or traits. There were 6,011 unique SNP-disease/trait combinations; as some SNPs have been associated with more than one disease or trait, these represent 5,386 unique dbSNP IDs. Of these, 5,654 associations and 5,134 SNPs were in noncoding regions (table S2). Coding regions were defined by the CCDS Project (downloaded from the UCSC genome browser at <http://hgdownload.cse.ucsc.edu/goldenPath/hg19/database/ccdsGene.txt.gz> on March 5, 2011) (37).

For some analyses, we grouped SNPs into classes of similar diseases or traits, namely, aging-related; autoimmune disease; cancer; cardiovascular diseases and traits; diabetes-related; drug metabolism; hematological; kidney, lung, or liver; lipids, miscellaneous, neurological/behavioral; parasitic or bacterial disease; quantitative traits; radiographic (primarily bone density); serum metabolites; and viral disease.

1.2. Identification of replicated GWAS associations.

Not all reported associations from GWAS studies are replicated when tested in subsequent studies of the same disease or trait. We examined whether associations with stronger evidence were more likely to map to DNase I hypersensitive sites (DHSs). Data in the GWAS catalog was tabulated and the SNPs divided into three overlapping classes (table S2) whose associations had varying levels of experimental support. SNPs were classified as "internally replicated" if the association was confirmed in a second replication population within the study as noted in the NHGRI GWAS Catalog. An association was classed as "externally replicated" if an association was observed in a second publication linking the same disease or trait to the same SNP. Associations which were not yet replicated by a second sample population within the study or by an independent study were classed as "unreplicated". A SNP could be included in both the "internally replicated" and "externally replicated" class; in such cases it was treated as externally replicated for the purpose of analysis.

2.1. DNase I mapping.

DNase I mapping was conducted on cultured cells, primary hematopoietic cells, and isolated fetal tissues using appropriate nuclei isolation protocols (table S1). Because the cell culture and isolation and handling protocols differ for different cell types, they are not included here but rather are all available online and indexed with URLs in table S1.

2.2. Isolation of nuclei from cultured cells.

Cells were grown in accordance with protocols obtained from the source (table S1). Freshly grown cells were centrifuged at 500g for 5 minutes (4°C) in an Eppendorf Centrifuge 5810R, and washed in cold PBS (Cellgro/Mediatech Inc.). Cell pellets were resuspended in Buffer A (15 mM Tris-Cl pH

8.0, 15 mM NaCl, 60 mM KCl, 1 mM EDTA (Ambion/Life Technologies Corp) pH 8.0, 0.5 mM EGTA (Boston BioProducts) pH 8.0, 0.5 mM spermidine (MP Biomedicals, LLC) and 0.15 mM spermine (MP Biomedicals, LLC) to a final concentration of 2×10^6 cells/mL. Nuclei were obtained by drop-wise addition of an equal volume of Buffer A containing 0.04% IGEPAL CA-630 (Sigma-Aldrich) to the cells, followed by incubation on ice for 10 min. Nuclei were centrifuged at 1,000g for 5 min and then resuspended and washed with 25 mL of cold Buffer A. Nuclei were resuspended in 2 mL of Buffer A at a final concentration of 1×10^7 nuclei/mL.

2.3. Isolation of nuclei from hematopoietic cells.

Lymphocyte subclasses were isolated by immunomagnetic separation. Cells were pelleted by centrifugation for 5 minutes at 500g at 4 °C. Cells were washed in ice-cold PBS, then resuspended to 5 million cells per mL in Buffer A. An equal volume of ice-cold 2X IGEPAL CA-630 solution (ranging from 0.02%-0.06%) was added and the tube was incubated for 5 - 6 minutes on ice to lyse the cells. Nuclei were pelleted by centrifugation for 5 minutes at 500g at 4 °C, resuspended in Buffer A and counted with a hemocytometer.

2.4. Isolation of nuclei from fetal tissues.

Tissue was minced, resuspended in cold 250 mM sucrose, 1 mM MgCl₂, 10 mM Tris-Cl pH 7.5, with added EDTA Protease Inhibitor Cocktail (Roche Applied Science Corp.). Resuspended tissue from fetal brain, fetal lung, fetal kidney, and fetal adrenal was dissociated by slowly homogenizing with a Dounce homogenizer. Resuspended tissue from fetal heart or fetal intestine was dissociated in a gentleMACS Dissociator (Miltenyi Biotech Inc.). Following dissociation, all fetal tissues were filtered through a 100 uM filter, and nuclei pelleted by centrifugation 600g for 10 minutes. Pelleted nuclei were washed with Buffer A, resuspended in Buffer A and counted in a hemocytometer.

2.5. DNase I mapping from isolated nuclei.

Briefly, DNase I digestion was performed as described in an earlier publication (15), with minor modifications. Isolated nuclei (2×10^6) from suspension cells or dissociated tissue were washed with 15 mM Tris-Cl pH 8.0, 15 mM NaCl, 60 mM KCl, 1 mM EDTA pH 8.0, 0.5 mM EGTA pH 8.0, 0.5 mM spermidine and 0.15 mM spermine then subjected to DNase I digestion for 3 min at 37 °C in 13.5 mM Tris-HCl pH 8.0, 87 mM NaCl, 54 mM KCl, 6 mM CaCl₂, 0.9 mM EDTA, 0.45 mM EGTA, 0.45 mM Spermidine. Digestion was stopped by addition of 50 mM Tris-HCl pH 8.0, 100 mM NaCl, 0.1% SDS, 100 mM EDTA pH 8.0, 1 mM spermidine, 0.3 mM spermine. A range of DNase I (Sigma-Aldrich), 10–80 U/mL concentrations was used for each preparation of nuclei and the sample giving the optimum difference between DNase I treated and untreated was used for sequencing library construction. DNase I double-hit fragments were collected by ultra-centrifugation and gel-purified. Adaptors were ligated to the ends of purified fragments, and the resulting libraries sequenced on an Illumina Genome Analyzer IIx according to a standard protocol.

2.6. Processing of DNase I-seq data.

For the ENCODE cell lines, the primary replicate was used for analysis. For the NIH Roadmap Epigenomics Consortium samples, data sets obtained from the tissues of fetal heart (12 developmental timepoint samples), fetal brain (12 developmental timepoint samples), fetal lung (34 developmental timepoint samples), fetal kidney (47 developmental timepoint samples), fetal intestine (15 developmental timepoint samples), fetal muscle (48 developmental timepoint and anatomical localization samples), fetal placenta (4 developmental timepoint samples), fetal skin (17 samples, 14 of which correspond to 7 replicate pairs from the same individual in different anatomical loca-

tions, 2 of which correspond to 1 replicate pair from a different individual and timepoint, and one sample from a third individual), fetal spinal cord (3 developmental timepoint samples), fetal stomach (11 developmental timepoint samples), fetal thymus (10 developmental timepoint samples), fetal adrenal (5 developmental timepoint samples), neonatal skin fibroblasts (4 samples corresponding to 2 replicate pairs from 2 different individuals), and neonatal skin keratinocytes (4 samples corresponding to 2 replicate pairs from 2 different individuals), we pooled the data following hotspot calculation from all timepoints and samples into a single DNase I hypersensitivity profile for each tissue. 36-base reads with up to two mismatches were mapped to the human genome (GRCh37/hg19) using the sequence aligner BOWTIE (38). DHSs were identified using the Hotspot algorithm (15) at a false discovery rate (FDR) threshold of 5%. Genomic feature overlaps and distance calculations were performed using the BEDOPS suite of software tools available at <http://code.google.com/p/bedops/> (39).

2.7. Data availability

The DNase I data used in this study have been released as part of the ENCODE Project (10) or the NIH Roadmap Epigenomics Mapping Consortium (11). Data released through both projects and available (table S1) include mapped reads and hotspots that have not been filtered for FDR thresholding. These data have been deposited in GEO under accession numbers GSE29692 and GSE18927. Data are also available for download through www.uwencode.org/data and through www.epigenomebrowser.org.

3.1. Enrichment of GWAS SNPs within DHSs relative to genomic space occupied.

The *P*-values for the enrichment of GWAS SNPs in DHSs, and various classes of DHSs, were computed using the binomial cumulative distribution function $b(x; n, p)$, the probability of x or more successes in n Bernoulli trials, with probability of success p . We used the R (40) function `pbinom` for calculating $b(x; n, p)$. We set the parameter n of the binomial equal to the total number of GWAS SNPs under consideration. For a given class of DHS we set the parameter p equal to the fraction of the 36-mer uniquely-mappable GRCh37/hg19 genome occupied by the DHS class (using 2,630,301,437 uniquely mappable bp), and parameter x equal to the number of the SNPs overlapped by the DHSs.

For comparison of the overlap of GWAS SNPs and DHSs to the overlap of HapMap (41) SNPs and DHSs, we obtained 4,029,798 CEPH population (Utah residents with ancestry from northern and western Europe, CEU) HapMap SNPs from the UCSC Genome Brower (release 27, merged Phase II + Phase III genotypes, lifted over from hg18 to hg19, downloaded from genome.ucsc.edu using the Table Brower). To compute the enrichment of GWAS SNPs in DHSs relative to the enrichment of HapMap SNPs in DHSs (fig. S1C), we set the expectation p equal to the fraction of HapMap SNPs overlapped by DHSs, we set n equal to the total number of GWAS SNPs, and we set x equal to the number of GWAS SNPs overlapped by DHSs.

3.2. Enrichment of GWAS SNPs in LD with SNPs in DHSs relative to randomly chosen 1KG SNPs.

We used CEU population genotype data from the 1000 Genomes Project (13) to compute the linkage disequilibrium (LD) measure r^2 between GWAS SNPs and SNPs in the DHSs near them. We converted the September 2010 release from GRCh36/hg18 to GRCh37/hg19 genomic coordinates using the UCSC Genome Brower liftOver tool. We then excluded SNPs for which a phased genotype was not available for all 60 CEU individuals sampled, or more than two alleles were present within the genotypes, or the minor allele frequency (MAF) was under 2/120. We then obtained the subset

of these that were GWAS SNPs lying within intronic and intergenic regions ($n = 4,885$), using the CCDS gene definitions (37). We computed r^2 between each such GWAS SNP lying outside a DHS and every SNP within a 125 kb radius lying within a DHS. We partitioned the overall results into three categories: GWAS SNPs within DHSs, GWAS SNPs achieving $r^2 = 1$ with a SNP lying within a DHS within a 125 kb radius, and all GWAS SNPs not belonging to the first two categories.

For each of 4,885 noncoding GWAS SNPs meeting our filtering criteria, we drew a SNP at random from the subset of 1000 Genomes noncoding SNPs having the same MAF, approximate distance from the transcription start site (TSS) of the nearest gene, and status of intronic or intergenic. This triple-matching procedure effectively accounts for any positional bias that might have been present in the SNP arrays. In addition to these three matching criteria, we also verified that the G+C content was the same between the GWAS SNPs and the matched control SNPs (table S3).

We obtained 1,000 independent, randomly-drawn replicate data sets of 4,885 SNPs, each set matched to the noncoding GWAS SNPs. For each replicate data set, we performed the r^2 calculations and categorization of results as we had done for the GWAS SNPs. We tallied the percentages of SNPs falling into these categories within each random data set and fit a normal distribution to these data (fig. S2). To estimate the P -value for observing as many of the GWAS SNPs as we did within the first two categories, we computed the area of the upper tail of this distribution that exceeded the percentage of GWAS SNPs falling into these categories (~78%). The upper tail had no detectable area in the range beyond 100%. The percentage of noncoding GWAS SNPs within DHSs or achieving $r^2 = 1$ with a SNP in a nearby DHS is significant at the level $P < 10^{-37}$.

To verify that the DHSs showing such strong associations with possibly-functional GWAS SNPs are not merely surrogates for coding exons, we then removed from consideration any DHS overlapping any coding exon by at least 1 bp, and re-measured the percentages of GWAS and random-matched SNPs falling within a DHS. This only removed ~4% of the DHSs, covering ~45 Mbp, from the pool, and hence had a negligible effect. ~77% of noncoding GWAS SNPs were found to lie within these DHSs or be in complete LD with them ($P < 10^{-28}$).

3.3. Calculation of F_{ST} for GWAS SNPs

We identified all noncoding autosomal sites for which 1000 Genomes (13) had fully-phased genotypes in both the CEU and Yoruba from Nigeria (YRI) populations, and partitioned these into sites within DHSs and sites outside of DHSs. We then chose 150,000 of these DHS sites at random, in the same proportion of intergenic to intronic sites that we observed in all noncoding 1000 Genomes CEU data across the autosomes (70.8% intergenic, 29.2% intronic). Next, for each intergenic DHS SNP, we chose an intergenic non-DHS SNP with the same minor allele frequency in CEU located at approximately the same distance from its nearest TSS, and did likewise for the intronic DHS SNPs. We filtered out any site at which the MAF pooled across the populations' genotypes fell below 10%, leaving us with 122,648 SNPs in the within-DHSs set and 122,810 SNPs in the non-DHS set. We computed F_{ST} and obtained values of 0.08433 and 0.08455 for these two SNP sets, respectively. Relaxing the restriction of matching on distance to the nearest TSS did not yield a significantly different result (0.08468). We also observed virtually no difference in F_{ST} between the two SNP sets when relaxing the constraint on MAF to 5% and 0%.

3.4. Disease-specific enrichment of GWAS SNPs in DHSs and fetal-origin DHSs

We computed the enrichment of GWAS SNPs from particular diseases or traits in DHSs (fig. S5) by dividing the proportion of GWAS SNPs in DHSs by the overall proportion of GWAS SNPs in DHSs (57.1%). Enrichments are reported as percentage enrichment or depletion. The individual significances of these enrichments was computed using the binomial distribution $b(x; n, p)$, setting the

parameter x to the number of GWAS SNPs of a given disease or trait in DHSs, n to the number of GWAS SNPs for the disease or trait, and p to 0.571.

We computed the enrichment of GWAS SNPs from particular diseases or traits in fetal-origin DHSs (Fig. 1E) by dividing the proportion of GWAS SNPs in fetal-origin DHSs by the overall proportion of GWAS SNPs in fetal-origin DHSs (88.1%). Enrichments are reported as percentage enrichment or depletion. The individual significances of these enrichments was computed using the binomial distribution $b(x; n, p)$, setting the parameter x to the number of GWAS SNPs of a given disease or trait in fetal-origin DHSs, n to the number of GWAS SNPs for the disease or trait, and p to 0.881. To compensate for the overall enrichment or depletion of disease categories in DHSs in general, GWAS SNPs not in any DHS were excluded.

4.1. DHS-to-promoter assignments based on cross-cell-type hypersensitivity correlations.

We previously measured DHSs genome-wide across 79 diverse cell types, and performed correlation analyses on the patterns of DNase I occupancy across the cell types (10). Briefly, we first collapsed the 79 cell types into 32 categories, based on the similarities and differences of their DHS profiles genome-wide (table S4). Then for each DHS, we formed a 32-element vector of DNase I tag counts to represent the occupancy pattern within those cell types at that DHS. Then for each promoter DHS representing a Gencode TSS, we computed the correlation between its occupancy pattern vector and the vector for each non-promoter DHS distal to it within a 500 kb radius. We defined a distal/promoter DHS pair to be “connected” if its Pearson correlation coefficient r was at least 0.7. We identified 578,905 connected distal DHSs genome-wide (mean separation = 266 kb), 429,283 (74%) of which hop over an adjacent gene to find its highest correlation with a different gene farther away within a 500-kb radius.

Here we used this correlation map to obtain a set of 296 unique noncoding GWAS SNPs lying within distal DHSs achieving $r \geq 0.7$ with a promoter DHS within 500 kb (table S6). We also repeated this analysis using DHSs found in 46 cell types that were used for other analyses in this paper but not included among the 79 used for the above (table S5). This correlation map identified an additional 123 unique noncoding GWAS SNPs lying within distal DHSs achieving $r \geq 0.7$ with a promoter DHS within 500 kb (table S7).

To establish the extent of LD between the distal and promoter DHSs, we computed r^2 between all pairs of 1000 Genomes SNPs fully phased in the CEU population and with minor allele frequency $\geq 5\%$ lying within 2 kb of the DHS containing the GWAS SNP and lying within 2 kb of the promoter DHS. For a typical DHS pair, we computed $\sim 127 r^2$ values, between ~ 14 SNPs at one DHS and ~ 9 SNPs at the other.

Two replicates of PolII ChIA-PET data (36) in K562 cells were obtained from the UCSC Genome Browser (<http://hgdownload-test.cse.ucsc.edu/goldenPath/hg19/encodeDCC/wgEncodeGisChiaPet/>) and processed with awk (42).

5.1. Transcription factor motif data.

Potential sites of transcription factor binding were identified by scanning relevant regions utilizing position weight matrices from three major transcription factor binding motif repositories: TRANSFAC (43), JASPAR (44), and UniPROBE (45). To avoid ascertainment bias for motifs better matching the reference allele of common polymorphisms, we created an alternate genome to complement the reference GRCh37/hg19 human genome. This alternate genome incorporates the non-reference allele at the location of each SNP identified in the CEU population of the 1000 Genomes Project (13).

Regions in the vicinity of GWAS or control SNPs were then scanned for motifs in both the reference and alternate genomes with a threshold $P \leq 10^{-4}$ using the program FIMO (46).

5.2. Mapping transcription factors to GWAS disease/trait classes.

We used information from the Gene Ontology (GO) (47) to identify potentially relevant motif matches. All GO biological processes for 282 transcription factors were extracted from the Gene Ontology MySQL database. For each disease/trait class, a collection of key terms which could identify factors potentially involved in the class was developed and used to search the list of GO biological processes associated with each transcription factor for which a position weight matrix was available (table S8). Many transcription factors were found to be consistent with multiple disease/trait classes. The set of transcription factor motifs detected ($P < 10^{-4}$), with at least one Gene Ontology Biological Process matching search terms for the disease/trait class and which overlapped GWAS SNPs in a DHS was identified and used for subsequent pathway/interaction analyses.

For our measurements of GWAS SNP enrichment within transcription factor motif groups, we began by forming a matrix of potential associations between transcription factor GO groups (e.g., aging) and disease classes (e.g., cancer). We computed the relative frequency with which GWAS SNPs from a particular disease class localized within the recognition sequence of a transcription factor annotated with related physiological processes, and derived a P -value using the binomial distribution $b(x; n, p)$, setting the first parameter to the number of GWAS SNPs present in the given factor group, and the second parameter to the proportion of GWAS SNPs belonging to the given disease class.

5.3. Allelic imbalance in chromatin accessibility.

We first called heterozygous SNPs directly from the DNase I reads. At each of the 5,386 unique GWAS SNPs (coding and noncoding), reads were extracted from DNase I alignments using SAMtools (48), and compared to the GRCh37/hg19 human reference sequence. To reduce the risk of false positives due to sequencing errors, only GWAS SNPs identified either in the 1000 Genomes Project's low-coverage CEU population data, or Complete Genomics' 54-individual sample were considered. To correct for mapping bias caused by the extra mismatch in reads containing the non-reference allele a less-stringent mismatch threshold was applied. Reads containing the reference allele were only counted if they contained zero or one base mismatches (over the entire read length) to the reference sequence; reads with the non-reference allele were counted if they had one or two base mismatches (one of which is the SNP). Any SNPs located within one read-length (36 bp) of another known SNP, represented by more than one chromosome in either sample from 1000 Genomes or Complete Genomics, were excluded from this analysis. Samples were called heterozygous at a SNP if each known allele was represented by reads aligned to at least three distinct positions (unique genomic coordinate and strand).

We identified 872 heterozygous SNPs, and pooled allele counts from all heterozygous samples. Confirming our strategy for avoiding reference mapping bias, we observed 412 SNPs with more reads from the reference allele, 416 SNPs with more reads containing the non-reference allele, and 44 SNPs with an equal amount of reads. Sites with fewer than 21 reads were excluded for lack of power to test for allelic imbalance. We then tested the remaining 584 sites for imbalance using a two-tailed binomial test. We calculated a false discovery rate using the R package qvalue (49). To set an overall cutoff for significantly imbalanced sites, we simulated 200 random sets of read counts at 584 sites using the binomial distribution, with the ratios at imbalanced sites sampled from the actual data. We tested the power of our method to correctly discover imbalanced sites, and measured the actual false discovery rate to be $< 5\%$ for a cutoff of $P < 0.025$.

6.1. Transcription factor-centered networks.

We obtained factors involved in maturity onset diabetes of the young (MODY, Fig. 3A), as well as those interacting with OTX1 (fig. S11), IRF9 (Fig. 3B), and ESRRA (fig. S12) using Ingenuity Pathways Analysis (Ingenuity Systems, www.ingenuity.com). The transcription factors in each network with known sequence specificities were examined for overlap with noncoding GWAS SNPs in DHSs in all cell types (the IRF9 network was restricted to cell types related to immune function: CD3+, CD3+_Cord_Blood, CD4+, CD8+, CD14+, CD19+, CD20+, CD34+, CD56+, fThymus, GM06990, GM12864, GM12865, GM12878, Th1, Th2, Th17, Jurkat). MODY factors were examined for GWAS SNPs associated with Type 1 or Type 2 diabetes or glucose metabolism-related traits. OTX1-interacting, IRF9-interacting, and ESRRA-interacting factors were examined for GWAS SNPs associated with neurological, autoimmune, and cancer classes, respectively. We tested the significance of the enrichment of disease-relevant GWAS SNPs in the TFs in these networks against the enrichment of random SNPs in the network. We used a binomial distribution with the parameter p set to the proportion of noncoding SNPs in the Affymetrix 500K genotyping array overlapping motifs of the same TFs in DHSs.

7.1. Disease networks.

For the autoimmune network (Fig. 4A), we used SNPs of the autoimmune class plus SNPs associated with Type 1 diabetes (table S2). We examined only GWAS SNPs in DHSs from cell types related to immune function and tallied the number of GWAS SNPs associated with autoimmune disease. Transcription factors overlapping 8 or more GWAS SNPs are shown.

For the cancer network (Fig. 4B), we used a set of GWAS SNPs associated with cancer in DHSs from all tissue types. Transcription factors overlapping 6 or more GWAS SNPs are shown.

For the psychiatric network (fig. S13), we used a set of GWAS SNPs associated with psychiatric diseases which were present in DHSs of fetal brain. Transcription factors overlapping 3 or more GWAS SNPs are shown, except for FOXI1 and FOXP3, which were removed from the network due to lack of hypersensitivity at their promoter DHSs.

For each network, we computed the significance of finding a set of TFs whose recognition sequences overlap such a high number of GWAS SNPs by comparing to random equally-sized samples of noncoding SNPs from the Affymetrix 500K genotyping array (10,000 replicates). P -values were estimated using a fitted Poisson distribution.

8.1. Cell type-selective GWAS variant-DHS enrichment analysis.

At a given P -value threshold, enrichment in a cell type's DHSs was calculated as the fraction of SNPs with a P -value below that threshold that overlap DHSs, divided by the fraction of all noncoding SNPs in the study that overlap DHSs. Malignancy-derived cell lines were excluded. Enrichments were tested at P -value thresholds from 1.0 to 10^{-75} . The thresholds were chosen as powers of ten which approximately halved the number of additional SNPs included at each successively-lower threshold. The smallest threshold was chosen to retain sufficient sample size (>100 SNPs). The statistical significance of each enrichment was measured with a one-sided Fisher's exact test, implemented in R's "fisher.test" function.

Supporting Figures

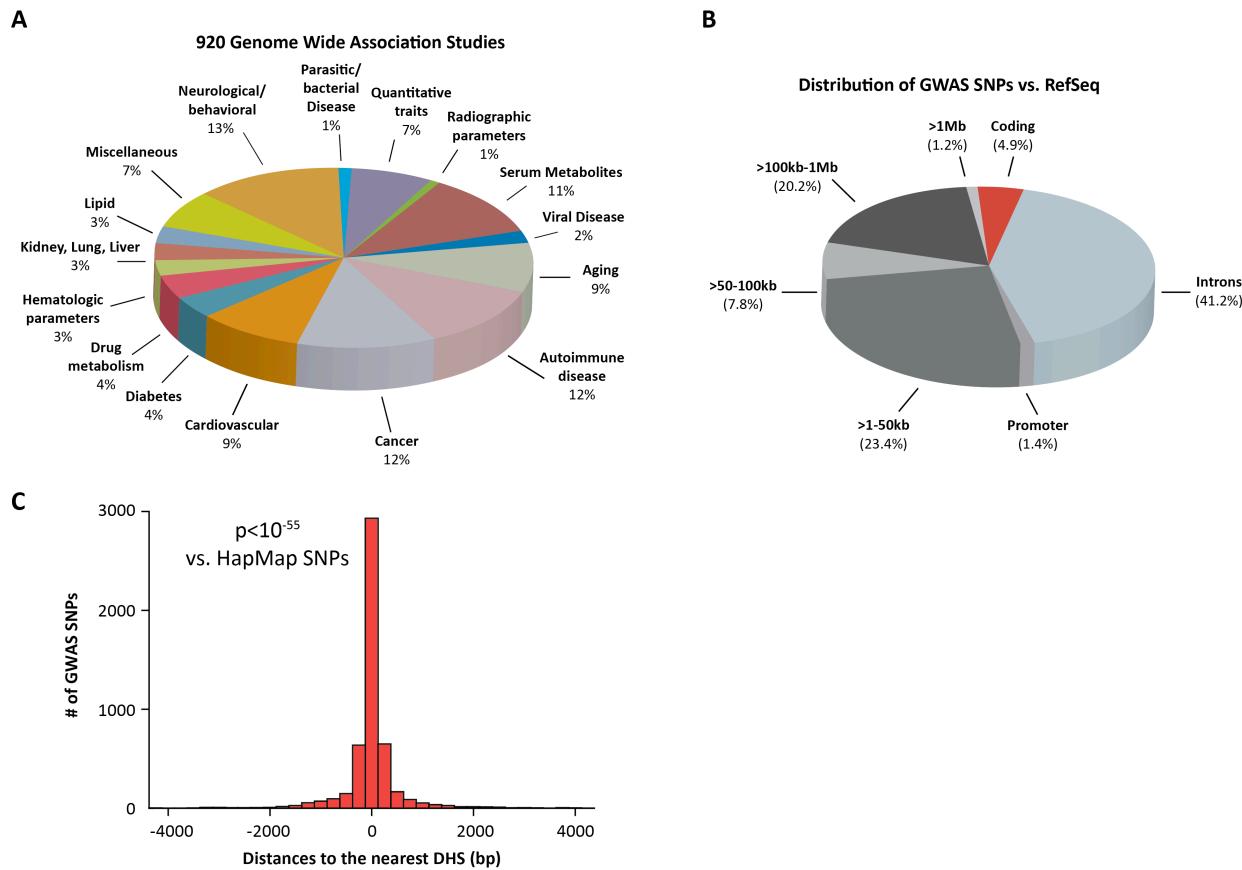


Fig. S1. Diseases and traits studied by GWAS and distribution of GWAS variants

(A) Catalog of 6,011 trait-SNP associations (5,386 distinct SNPs) from 920 different studies. Chart shows percentage of GWAS SNPs by disease/trait class.

(B) Location of GWAS SNPs relative to genic features. Note only 4.9% of GWAS SNPs lie in coding sequence.

(C) Overlap of noncoding GWAS SNPs (5,134 distinct SNPs) and regulatory DNA. Horizontal axis: Binned distances from DHSs. Central "0" bin contains only GWAS SNPs within DHSs. The overlap is highly significant, even when corrected for a baseline enrichment of HapMap SNPs in DHSs.

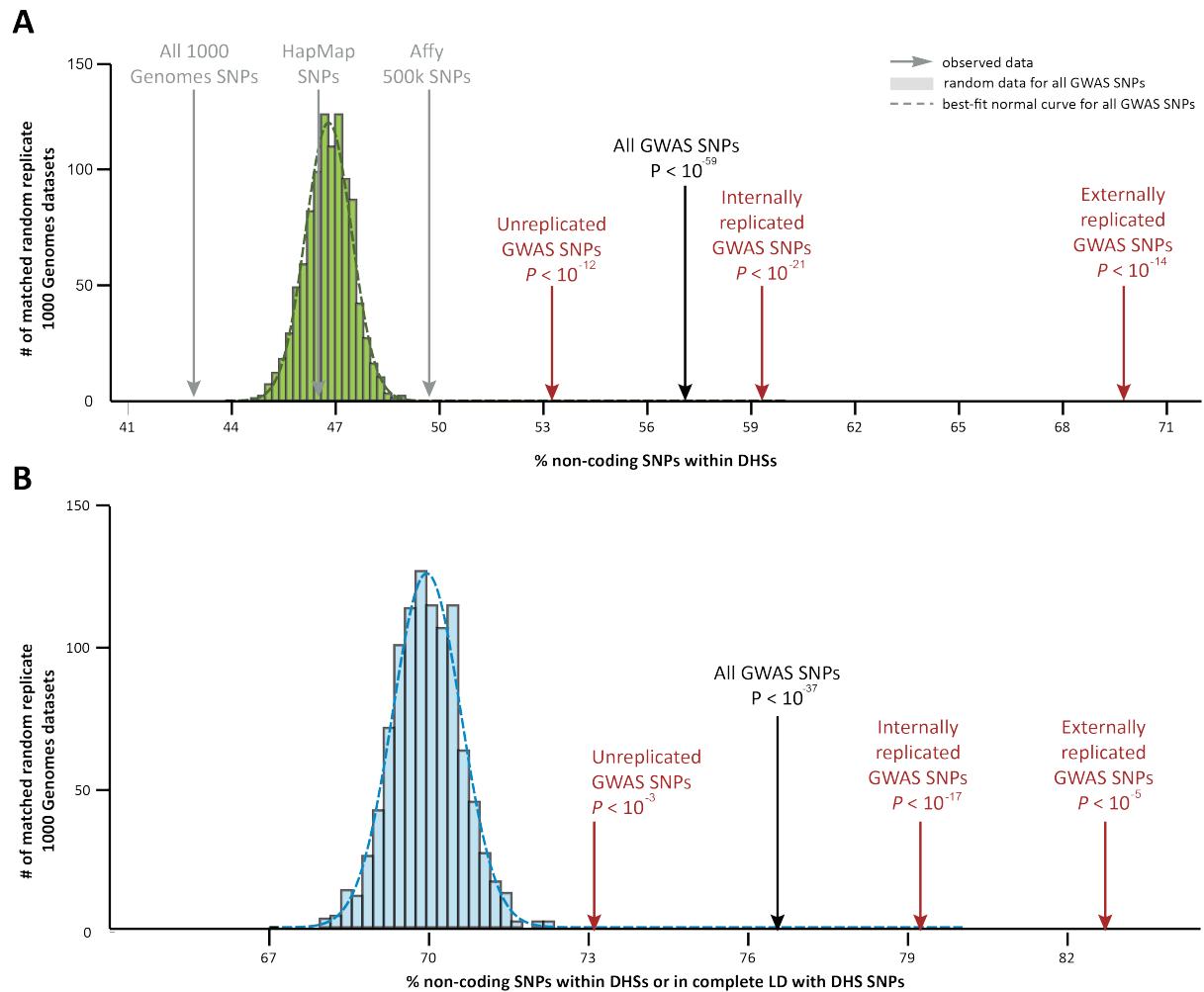


Fig. S2. Overlap of noncoding GWAS SNPs and regulatory DNA marked by DHSs.

(A) Best-fit normal distribution of 1000 independent replicates of randomly-sampled SNPs matching all noncoding GWAS SNPs in genomic feature localization (intronic vs. intergenic), distance from the nearest TSS, and MAF in northwestern European populations; P -value (black arrow) was estimated as the area under the corresponding portion of the tail. Note the monotonic increase in the enrichment of disease/trait variants in DHSs with increasing quality of GWAS SNP experimental replication (red arrows); P -values shown in red were estimated from distributions (not shown) matched to each class of SNPs. Control sets consisting of all noncoding 1000 Genomes, HapMap CEU SNPs and Affymetrix 500K SNPs are shown for comparison.

(B) Same as (A), but measuring the percentage of GWAS SNPs within or in complete LD with 1000 Genomes SNPs in DHSs.

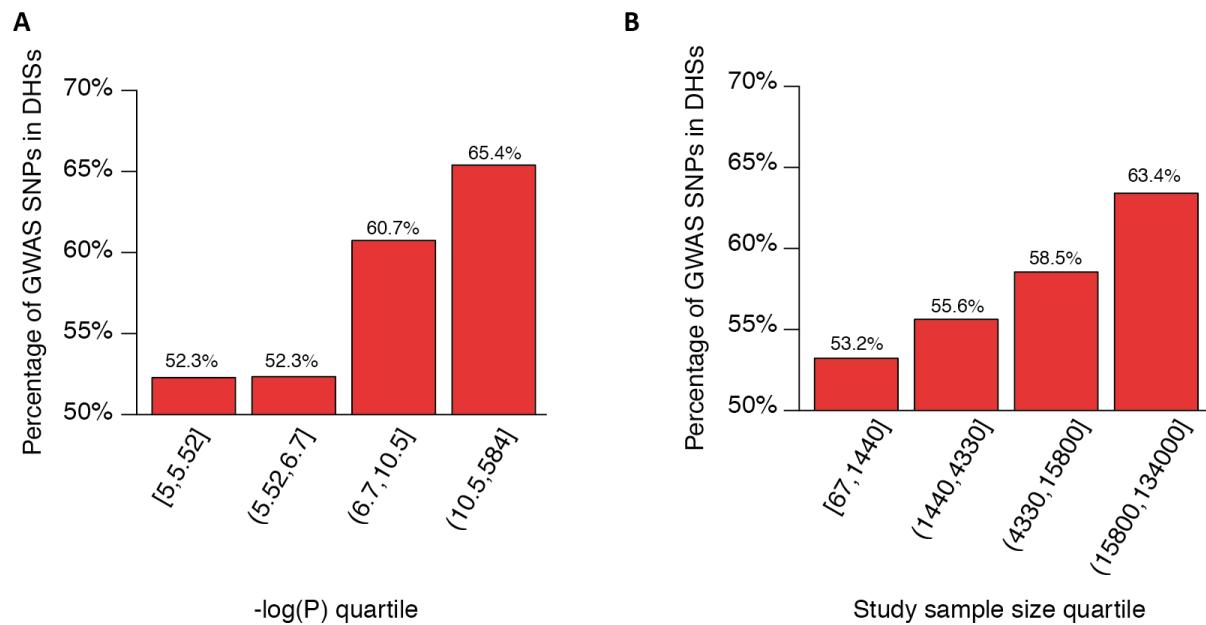


Fig. S3. Enrichment for regulatory DNA increases with strength of association.

Overlap of GWAS SNPs in specified P -value (A) and sample size (B) quartiles with DHSs. Note the progressive enrichments parallel Fig. 1B.

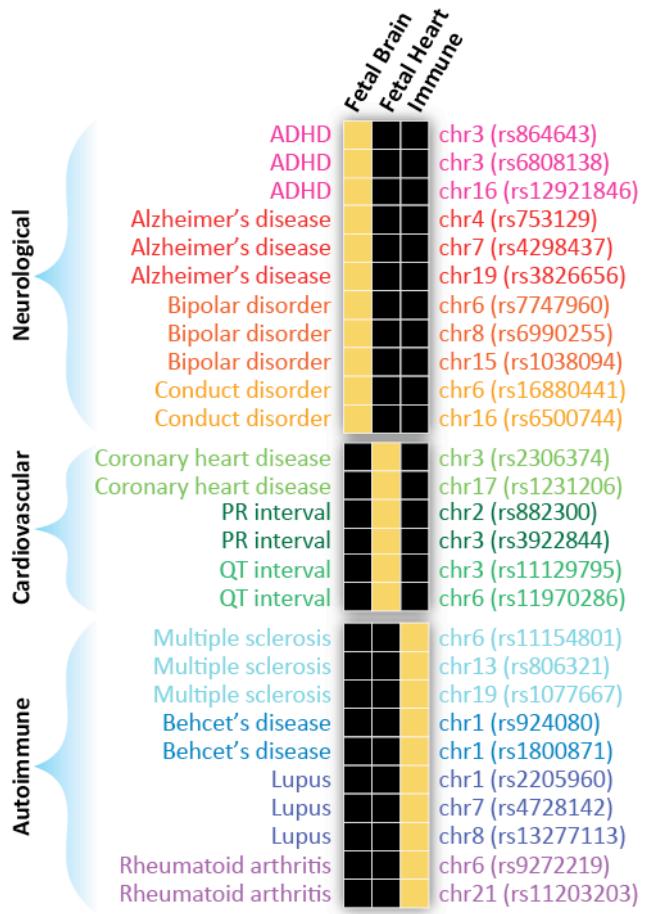


Fig. S4. Multiple distinct genomic disease associations repeatedly localize within relevant cell-selective DHSs.

Each cell represents the presence or absence of a DHS at the location of the given GWAS SNP. Yellow = DHS present in that cell/tissue class; black = absent.

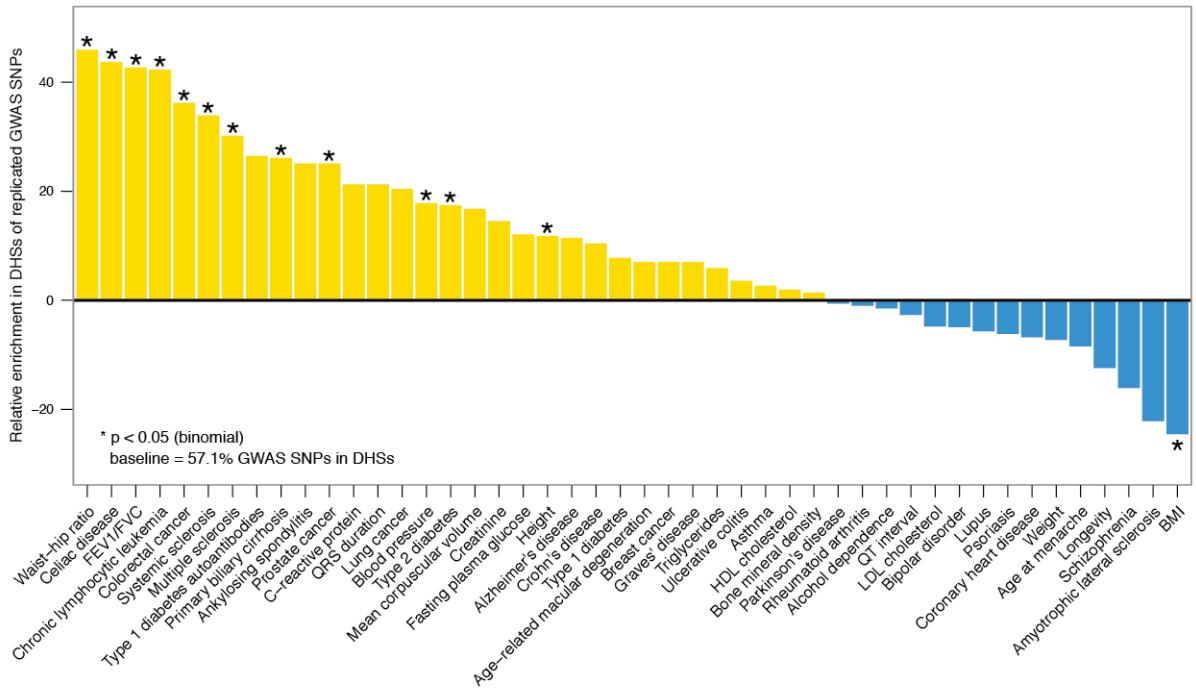


Fig. S5. Enrichment of GWAS SNPs for DHSs by disease/trait.

Shows the magnitude of enrichment or depletion of replicated GWAS SNPs in DHSs (Y-axis) for a given disease/trait (X-axis), relative to the background prevalence of all GWAS SNPs in DHSs (57.1%). Asterisks indicate the significance of the enrichment ($P < 0.05$, binomial). Only traits with >15 internally- or externally-replicated associations are shown.

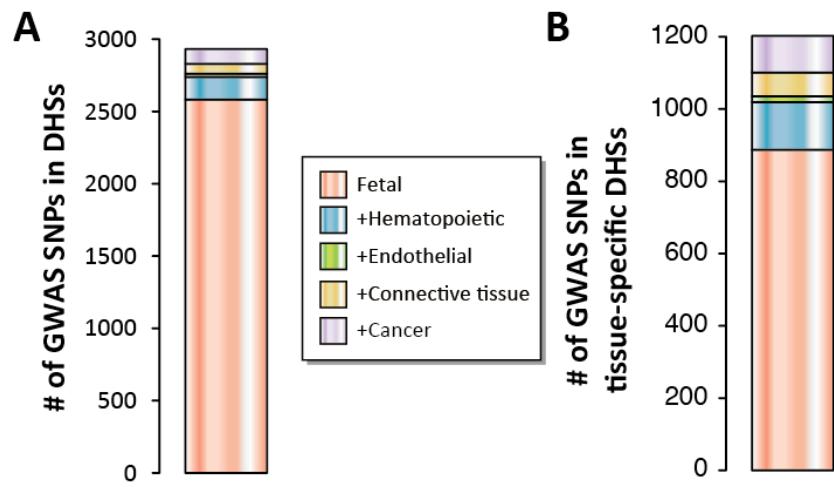


Fig. S6. Localization of GWAS SNPs in DHSs of fetal and adult tissue classes.

(A) Cumulative tally of GWAS SNPs by DHS tissue category. Each color denotes SNPs overlapping DHSs in that tissue type but not in preceding categories. Note that the vast majority of adult-stage DHSs with GWAS variants derive from either differentiated hematopoietic cells or cancer lines.

(B) Same as (A) except for DHSs specific to a tissue class.

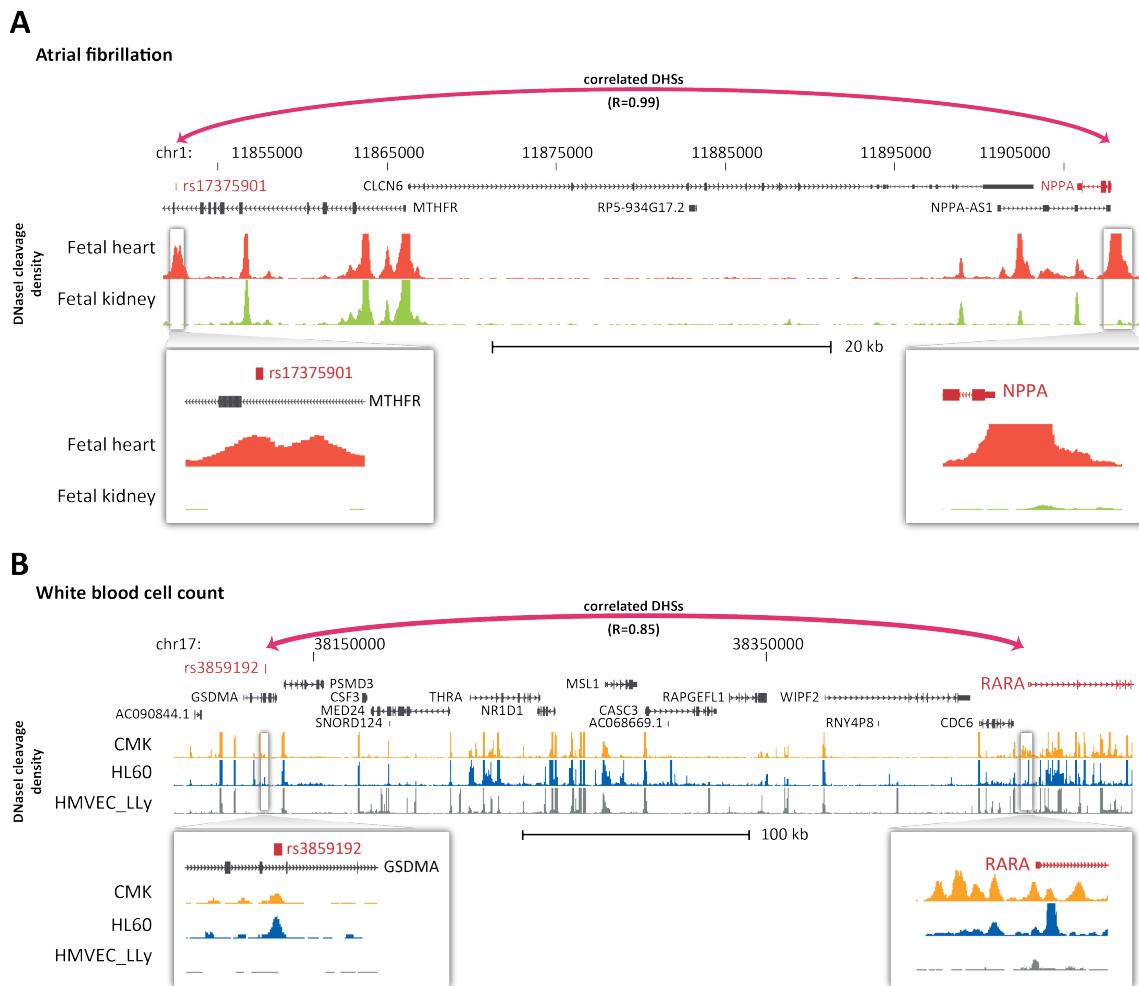


Fig. S7. Regulatory GWAS variants are linked to distant target genes.

(A) A DHS specific to fetal heart, connected with the gene encoding atrial natriuretic peptide A (NPPA), harbors a GWAS variant associated with atrial fibrillation.

(B) Distant (336 kb) DHS connected with retinoic acid receptor-alpha (RARA), a nuclear receptor involved in myeloid differentiation, harbors a GWAS variant associated with white blood cell count.

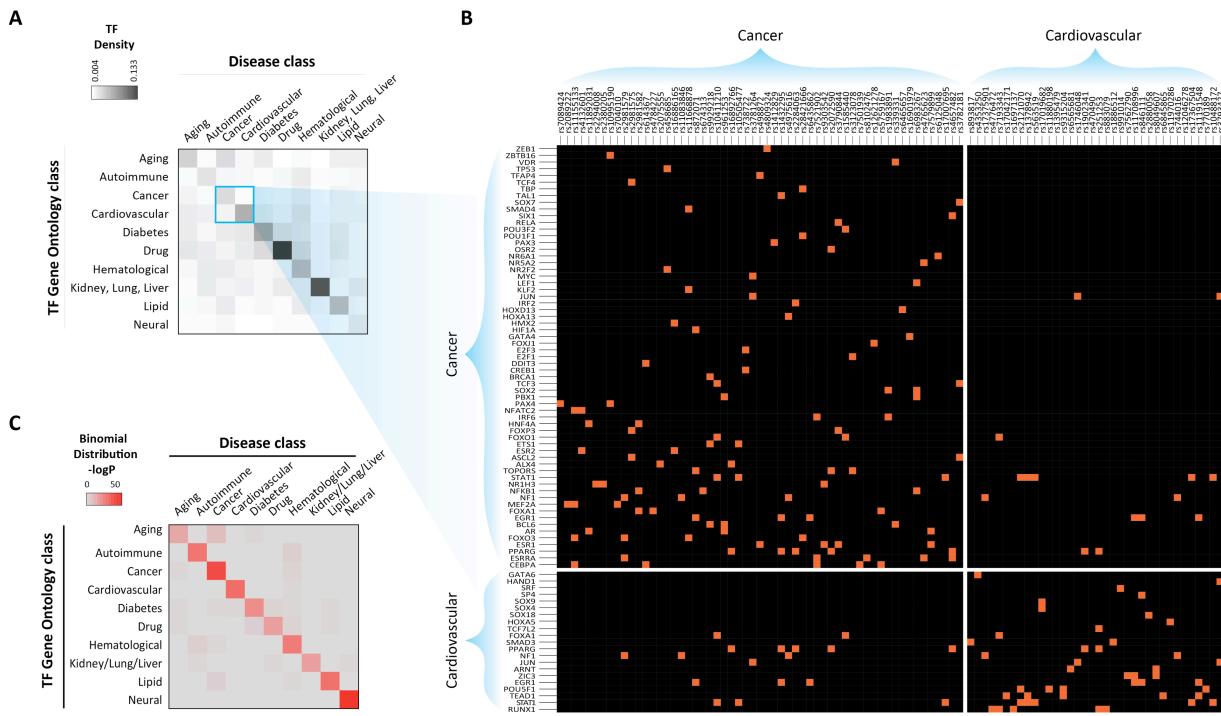


Fig. S8. GWAS variants in DHSs localize within physiologically relevant TF binding sites.

(A) GWAS variants in DHSs localize within physiologically relevant TF binding sites. Columns: Categorization of GWAS SNPs by disease area (table S2). Rows: Transcription factor categories from unbiased mapping into disease/pathophysiologic categories with GO. Each cell shows the proportion (grayscale bar) of all GWAS SNPs from a given disease category (column) that fall into the binding sites of TFs within each TF category (row).

(B) Close-up of cancer and cardiovascular diseases showing the presence (red) or absence (black) of a recognition sequence for a particular recognition sequence (rows) at the location of a GWAS SNP in the indicated disease category (columns).

(C) Significance of proportions in (A), with high significance (binomial test) along the diagonal indicating systematic localization of GWAS variants from a given disease category within binding sites of pathophysiologically-related TFs

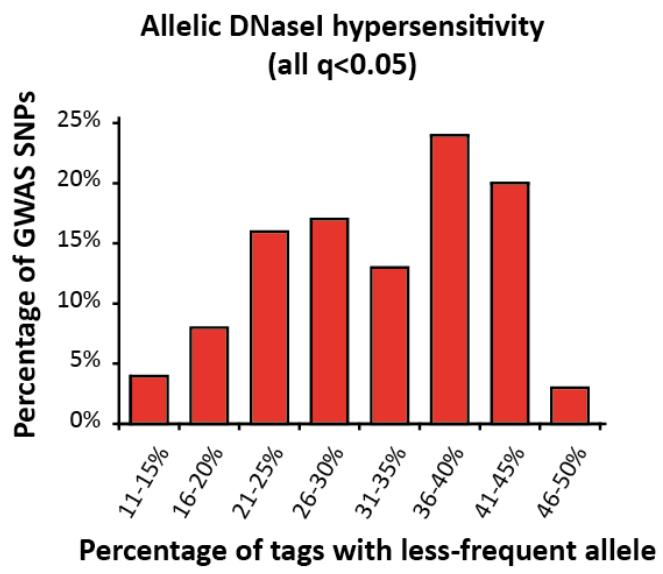


Fig. S9. Allelic imbalance distribution.

Distribution of the proportion of reads from the less-frequent allele at DHSs with significant (FDR < 5%) imbalance in DNase I hypersensitivity.

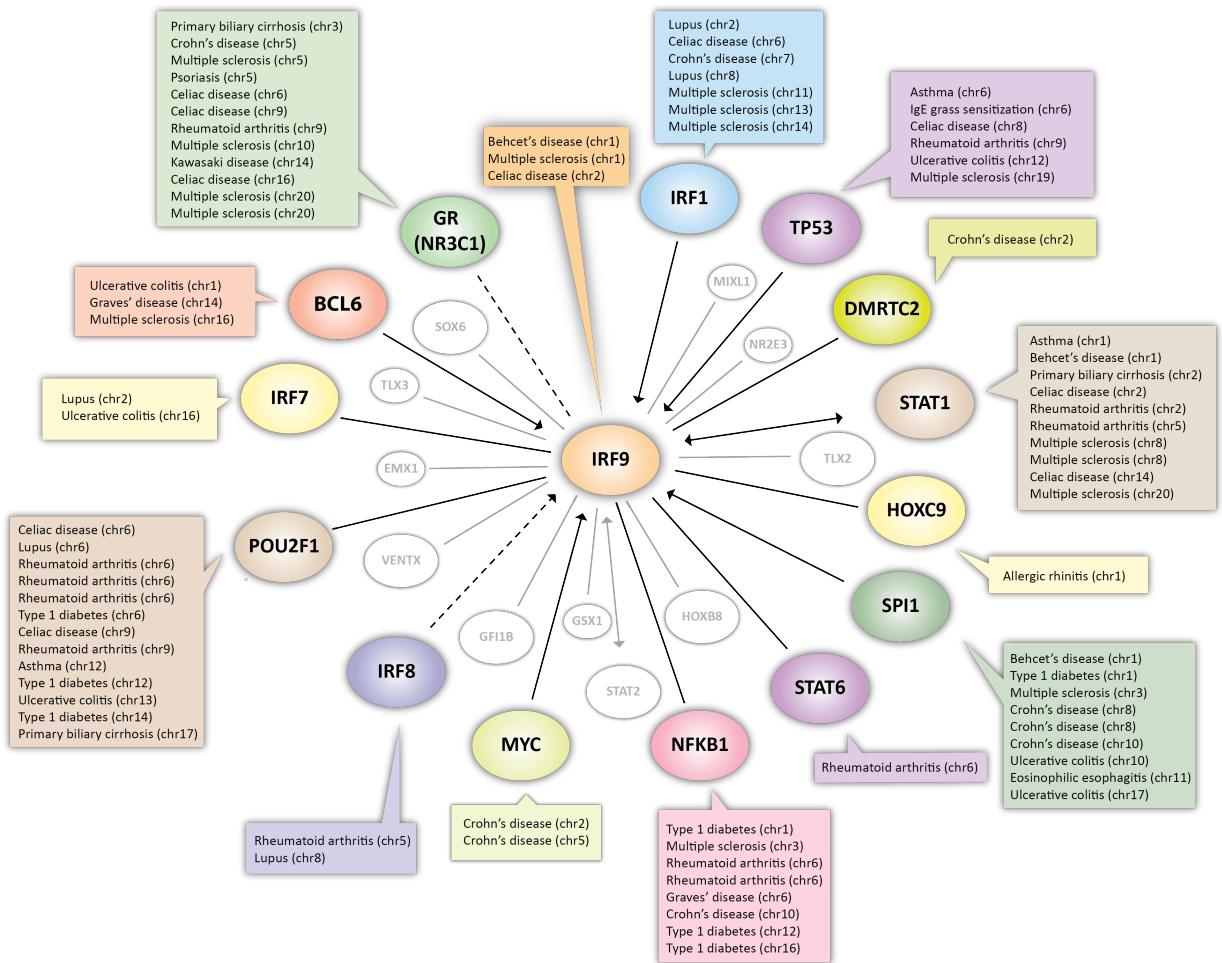


Fig. S10. Autoimmune disease-associated SNPs cluster in IRF9 interaction network (full network).

Detail of Fig. 3B showing factors for which no position weight matrices were available (small gray ellipses, n=6) or for which no GWAS SNPs were in their binding sites (large gray ellipses, n=5). SNPs in DHSs associated with autoimmune diseases repeatedly localize in recognition sequences for transcriptional regulators (labeled ellipses) that interact with IRF9. Arrows indicate directionality of relationship; dotted lines represent indirect interactions (12).

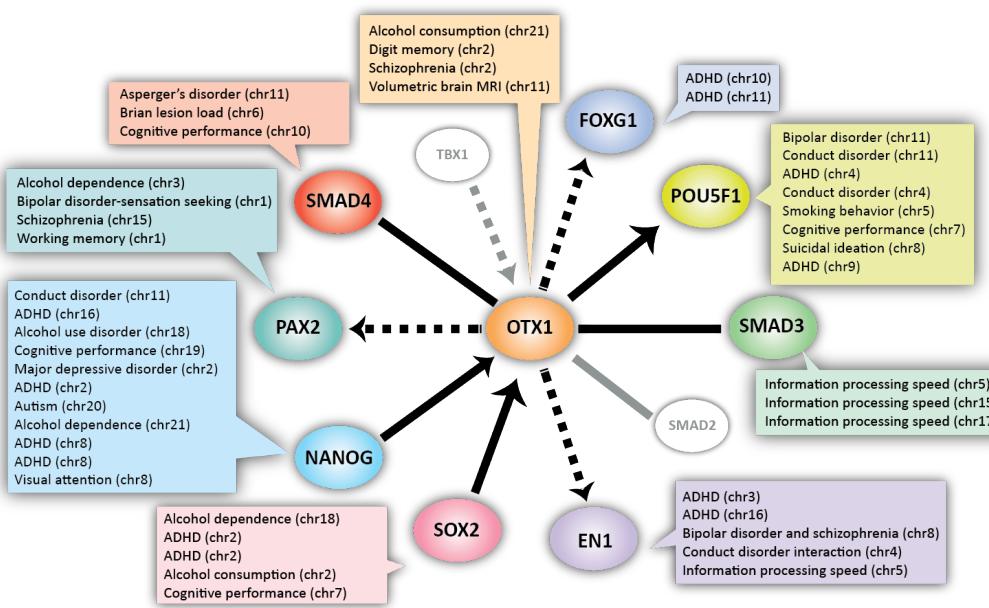


Fig. S11. Repeated involvement of the OTX1 pathway in neuropsychiatric diseases and traits.

SNPs in DHSs associated with diverse neuropsychiatric diseases and traits repeatedly localize within recognition sequences of TFs that interact with the brain morphogenic factor OTX1. SNPs in DHSs associated with cancer repeatedly localize in recognition sequences for transcriptional regulators (labeled ellipses) that interact with ESRRA, significant at $P < 0.049$ (binomial; 1.3x enrichment vs. proportion of random SNPs) (12). Arrows indicate directionality of relationship; dotted lines represent indirect interactions (12). Large gray ellipses indicate factors for which position weight matrices were available, but no recognition sequences overlapped GWAS SNPs.

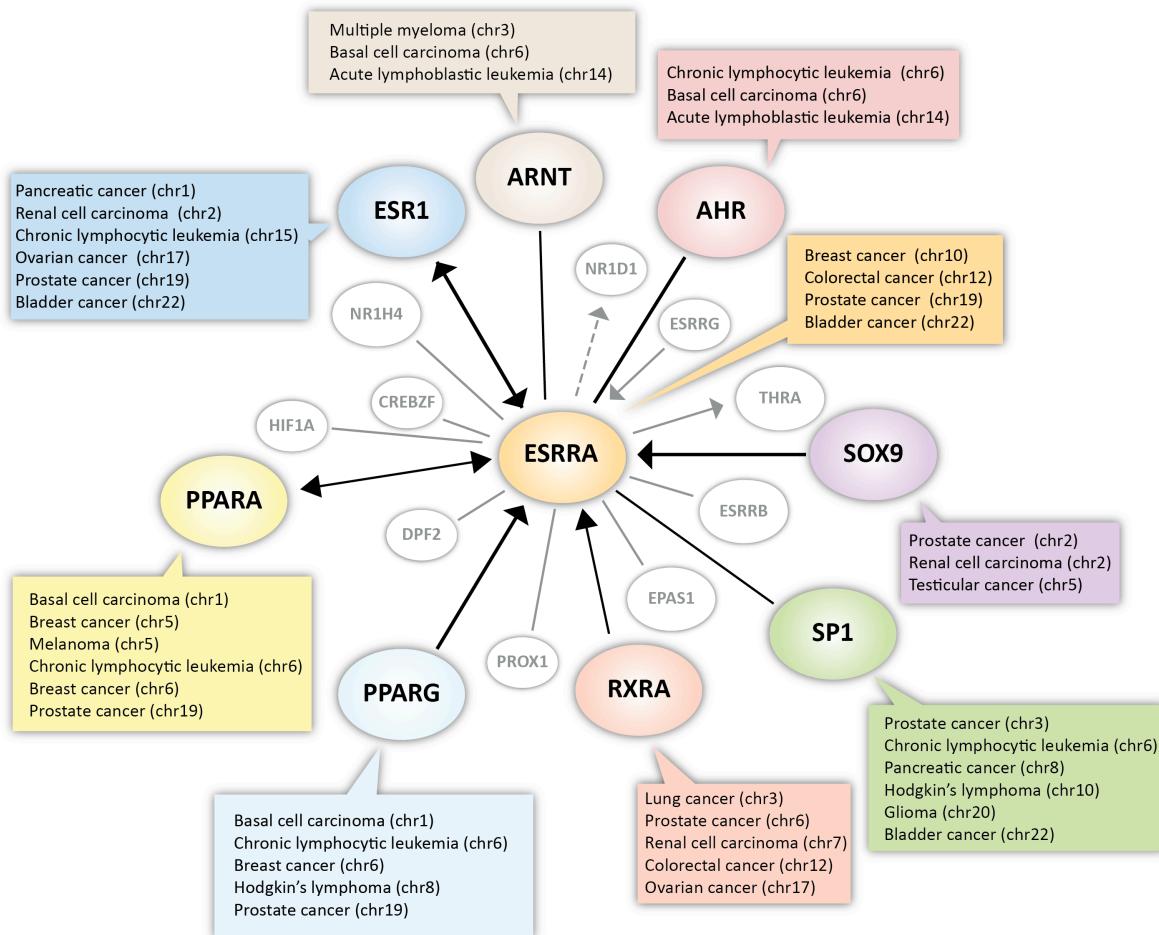


Fig. S12. Cancer-associated SNPs cluster in orphan nuclear receptor ESRRA network.

SNPs in DHSs associated with cancer repeatedly localize in recognition sequences for transcriptional regulators (labeled ellipses) that interact with ESRRA, significant at $P < 0.010$ (binomial; 1.5x enrichment vs. proportion of random SNPs) (12). Arrows indicate directionality of relationship, dotted lines represent indirect interactions (12). Large gray ellipses indicate factors for which position weight matrices were available, but no recognition sequences overlapped GWAS SNPs; small gray ellipses indicate that a position weight matrix for this factor is unavailable. The 28 distinct SNPs in the ESRRA network represent 12.7% of 220 cancer GWAS SNPs overlapping DHSs.

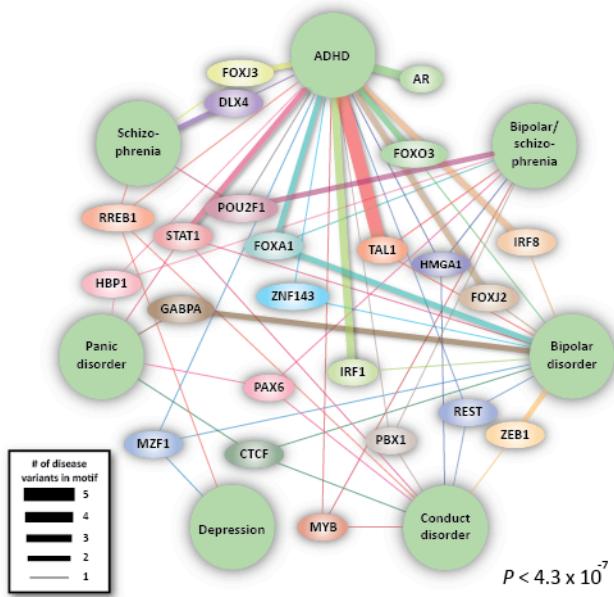


Fig. S13. Psychiatric disease network.

GWAS SNPs from related diseases repeatedly perturb recognition sequences of common transcription factors. Shown are factors whose recognition sequences harbor ≥ 3 GWAS SNPs in psychiatric diseases. Edge thickness represents number of associations between TF and disease in DHSs in relevant tissues.

Supporting Tables

Table S1. Mapping of DHSs in 349 cell and tissue samples.

DNase I mapping of 349 cell types and tissues (115 distinct types) used in the study, including the shorthand name for the tissue, a description of the tissue, whether the tissue is of fetal origin, the total number of DHSs observed, the number of GWAS SNPs within the DHSs, whether the DNase I data has been previously published in (10), and the preparation protocol for the cell line or tissue.

Cell_line	Description	Fetal?	#DHSs	#SNPs	Pub?	Cell/Tissue Isolation/Culture Protocol
A549	Epithelial cell line derived from a lung carcinoma tissue	N	117,992	180	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/A549_Stam_protocol.pdf
AG04449	Fetal buttock/thigh fibroblast	Y	174,802	202	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/AG04449_Stam_protocol.pdf
AG04450	Fetal lung fibroblast	Y	150,114	187	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/AG04450_Stam_protocol.pdf
AG09309	Adult human toe fibroblast	N	197,301	266	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/AG09309_Stam_protocol.pdf
AG09319	Adult human gum tissue fibroblasts	N	137,192	190	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/AG09319_Stam_protocol.pdf
AG10803	Adult human abdominal skin fibroblasts	N	171,903	224	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/AG10803_Stam_protocol.pdf
AoAF	Normal Human Aortic Adventitial Fibroblast Cells	N	169,477	261	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/AoAF_Stam_protocol.pdf
BE2_C	Human Neuroblastoma cell line	N	168,003	259	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/BE2_C_Stam_protocol.pdf
BJ	Skin fibroblasts	N	162,671	246	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/BJ-tert_Stam_protocol.pdf
Caco-2	Colorectal adenocarcinoma	N	117,293	179	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/Stam_15_protocols.pdf
CD14+	Monocytes, CD14+	N	117,181	273	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/Mono_CD14_Stam_protocol.pdf
CD19+	B-lymphocytes, CD19+	N	75,086	225	N	roadmapepigenomics.org/files/protocols/experimental/dnase1-sensitivity/HematopoieticCells_DNaseTreatment_V5_UW-NREMC.pdf
CD20+	B-lymphocytes, CD20+	N	170,412	328	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/CD20+_Stam_protocol.pdf
CD20+	B-lymphocytes, CD20+	N	86,908	268	N	genome.ucsc.edu/ENCODE/protocols/cell/human/CD20+_Stam_protocol.pdf
CD3+	T-lymphocytes, CD3+	N	77,933	177	N	roadmapepigenomics.org/files/protocols/experimental/dnase1-sensitivity/HematopoieticCells_DNaseTreatment_V5_UW-NREMC.pdf
CD34+	Mobilized hematopoietic progenitor cells	N	134,718	230	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/CD34+Mobilized_Stam_protocol.pdf
CD3+_Cor_dBlood	Cord blood, CD3+	N	74,992	176	N	roadmapepigenomics.org/files/protocols/experimental/dnase1-sensitivity/HematopoieticCells_DNaseTreatment_V5_UW-NREMC.pdf
CD4+	T helper cells, CD4+	N	94,881	239	N	roadmapepigenomics.org/files/protocols/experimental/dnase1-sensitivity/HematopoieticCells_DNaseTreatment_V5_UW-NREMC.pdf
CD56+	Lymphocytes, CD56+	N	105,724	277	N	roadmapepigenomics.org/files/protocols/experimental/dnase1-sensitivity/HematopoieticCells_DNaseTreatment_V5_UW-NREMC.pdf
CD8+	Cytotoxic T cells, CD8+	N	75,382	185	N	roadmapepigenomics.org/files/protocols/experimental/dnase1-sensitivity/HematopoieticCells_DNaseTreatment_V5_UW-NREMC.pdf
CMK	Acute megakaryocytic leukemia cells	N	123,561	210	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/CMK_Stam_protocol.pdf

Cell_line	Description	Fetal?	#DHSs	#SNPs	Pub?	Cell/Tissue Isolation/Culture Protocol
GM06990	Lymphoblastoid	N	86,958	210	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/Stam_15_protocols.pdf
GM12864	Lymphoblastoid	N	132,370	262	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/GM12864_Stam_protocol.pdf
GM12865	Lymphoblastoid	N	133,962	280	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/GM12865_Stam_protocol.pdf
GM12878	Lymphoblastoid	N	109,419	240	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/Stam_15_protocols.pdf
H1_P18	H1-derived embryonic stem cells	-	178,572	255	N	Yu et al., Cell Stem Cell 8, 326-334 (2011)
H7-hESC	Undifferentiated embryonic stem cells	-	284,627	305	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/H7-hESC_Stam_protocol.pdf
H9_P42	H9-derived embryonic stem cells	-	140,166	192	N	Yu et al., Cell Stem Cell 8, 326-334 (2011)
HAEpiC	Human Amniotic Epithelial Cells	Y	200,771	292	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/HAEpiC_Stam_protocol.pdf
HAc	Human Astrocytes - cerebellar	Y	183,752	239	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/HAc_Stam_protocol.pdf
HAh	Human Astrocytes - hippocampal	Y	215,151	351	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/HAh_Stam_protocol.pdf
HAsp	Human Astrocytes - Spinal cord	Y	215,720	350	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/HA-sp_Stam_protocol.pdf
HBMEC	Human Brain Microvascular Endothelial Cells	Y	196,870	320	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/HBMEC_Stam_protocol.pdf
HCF	Human Cardiac Fibroblasts	Y	171,858	268	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/HCF_Stam_protocol.pdf
HCFAa	Human Cardiac Fibroblasts - adult atrial	N	184,810	323	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/HCFAa_Stam_protocol.pdf
HCM	Human Cardiomyocytes	Y	191,262	308	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/HCM_Stam_protocol.pdf
HCPEpiC	Human Choroid Plexus Epithelial Cells	Y	209,492	304	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/HCPEpiC_Stam_protocol.pdf
HCT-116	Colon adenocarcinoma	N	104,196	170	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/HCT116_Stam_protocol.pdf
HConF	Human Conjunctival Fibroblasts	Y	150,877	209	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/HConF_Stam_protocol.pdf
HEEpiC	Human Esophageal Epithelial Cells	Y	213,954	266	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/HEEpiC_Stam_protocol.pdf
HepG2	Hepatocellular carcinoma	N	81,159	133	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/Stam_15_protocols.pdf
HESC	Human H1 Embryonic Stem Cell line	-	163,880	195	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/HHSEC_Stam_protocol.pdf
HFF	Human Foreskin Fibroblasts	N	189,148	329	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/HFF_Stam_protocol.pdf
HFF_Myc	Human Foreskin Fibroblasts_Myc Transgene	N	215,171	333	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/HFFMyc_Stam_protocol.pdf
HGF	Human Gingival Fibroblasts	N	148,852	191	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/HGF_Stam_protocol.pdf
HIPEpiC	Human Iris Pigment Epithelial Cells	Y	231,963	304	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/HIPEpiC_Stam_protocol.pdf
HL-60	Human promyelocytic leukemia cells	N	153,865	296	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/HL-60_Stam_protocol.pdf
HMEC	Human mammary epithelial cells	N	139,620	214	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/HMEC_Stam_protocol.pdf
HMF	Human Mammary Fibroblasts	N	176,102	236	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/HMF_Stam_protocol.pdf
HMVEC-LBI	Human Lung Blood Microvascular Endothelial Cells	N	161,548	283	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/HMVECLBI_Stam_protocol.pdf
HMVEC-LLy	Human Lung Lymphatic Microvascular Endothelial Cells	N	130,544	235	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/HMVECLLy_Stam_protocol.pdf
HMVEC-dAd	Adult Human Dermal Microvascular Endothelial Cells	N	115,973	175	N	genome.ucsc.edu/ENCODE/protocols/cell/human/HMVECdAd_Stam_protocol.pdf
HMVEC-dBl-Ad	Adult Human Dermal Blood Microvascular Endothelial Cells	N	149,796	268	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/HMVECdBl-Ad_Stam_protocol.pdf
HMVEC-dBl-Neo	Neonatal Human Dermal Blood Microvascular Endothelial Cells	N	154,291	310	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/HMVECdBl-Neo_Stam_protocol.pdf
HMVEC-dLy-Ad	Adult Human Dermal Lymphatic Microvascular Endothelial Cells	N	115,834	194	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/HMVECdLy-Ad_Stam_protocol.pdf
HMVEC-	Neonatal Human Dermal	N	139,708	242	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/HMVE

Cell_line	Description	Fetal?	#DHSs	#SNPs	Pub?	Cell/Tissue Isolation/Culture Protocol
dLy-Neo	Lymphatic Microvascular Endothelial Cells					C-dLy-Neo_Stam_protocol.pdf
HMVEC-dNeo	Neonatal Human Dermal Microvascular Endothelial Cells	N	132,325	215	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/HMVEC-dNeo_Stam_protocol.pdf
HNPC-EpiC	Human Non-Pigment Ciliary Epithelial Cells	Y	217,558	296	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/HNPEC_EpiC_Stam_protocol.pdf
HPAEC	Human Pulmonary Artery Endothelial Cells	N	125,462	170	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/HPAEC_Stam_protocol.pdf
HPAF	Human Pulmonary Artery Fibroblasts	Y	181,244	302	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/HPAF_Stam_protocol.pdf
HPF	Human Pulmonary Fibroblasts	Y	147,153	225	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/HPF_Stam_protocol.pdf
HPdLF	Human Periodontal Ligament Fibroblasts	N	169,679	260	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/HPdLF_Stam_protocol.pdf
HRCEpiC	Human renal cortical epithelial cells (normal)	N	193,462	294	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/HRCEpiC_Stam_protocol.pdf
HRE	Human renal epithelial cells (normal)	N	197,779	257	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/HRE_Stam_protocol.pdf
HRGEC	Human Renal Glomerular Endothelial Cells	Y	143,319	188	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/HRGE_C_Stam_protocol.pdf
HRPEpiC	Human Retinal Pigment Epithelial Cells	Y	229,606	298	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/HRPEpiC_Stam_protocol.pdf
HSMM	Human Skeletal Muscle Myoblasts	N	234,182	335	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/HSMM_Stam_protocol.pdf
HSMM_D	Human Skeletal Muscle Myoblasts_differentiated	N	233,756	414	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/HSMM_Stam_protocol.pdf
HUVEC	Human umbilical vein endothelial cells	N	115,081	229	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/Stam_15_protocols.pdf
HVMF	Human Villous Mesenchymal Fibroblasts	Y	170,308	296	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/HVMF_Stam_protocol.pdf
HeLa-S3	Cervical carcinoma	N	119,081	247	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/Stam_15_protocols.pdf
IMR90	Fibroblasts	N	196,940	278	N	genome.ucsc.edu/ENCODE/protocols/cell/human/IMR90_Stam_protocol.pdf
Jurkat	T lymphoblastoid cell line derived from acute T cell leukemia	N	152,487	251	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/Jurkat_Stam_protocol.pdf
K562	Chronic myeloid leukemia	N	142,920	268	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/Stam_15_protocols.pdf
LNCaP	Prostate adenocarcinoma cell line	N	184,899	239	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/LNCaP_Stam_protocol.pdf
MCF-7	Mammary gland adenocarcinoma	N	133,229	168	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/Stam_15_protocols.pdf
Mesendoderm	H1 derived mesendoderm cells	-	214,950	273	N	Vodyanik et al., Cell Stem Cell 7, 718-729 (2010)
NB4	Acute Promyelocytic Leukemia cell line	N	131,948	240	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/NB4_Stam_protocol.pdf
NH-A	Normal Human Astrocytes	Y	189,150	280	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/NHA_Stam_protocol.pdf
NHDF-Ad	Adult Human Dermal Fibroblasts	N	226,683	330	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/NHDF-Ad_Stam_protocol.pdf
NHDF-neo	Neonatal Human Dermal Fibroblasts	N	184,888	269	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/NHDF-neo_Stam_protocol.pdf
NHEK	Normal Human Epidermal Keratinocytes	N	145,886	216	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/Stam_15_protocols.pdf
NHLF	Normal Human Lung Fibroblasts	N	204,839	296	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/NHLF_Stam_protocol.pdf
NPC	H1 derived neuroprogenitor cells	-	93,396	148	N	N/A
NT2-D1	Human malignant pluripotent embryonal cancer cell line - Induced by RA to neuronal cells	N	187,959	259	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/Stam_15_protocols.pdf
PANC-1	Pancreatic carcinoma cell line	N	117,169	203	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/PANC-1_Stam_protocol.pdf
PrEC	Human Prostate Epithelial Cell Line	N	176,183	220	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/PrEC_Stam_protocol.pdf
RPTEC	Human Renal Proximal Tubule Cells	N	171,601	293	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/RPTEC_Stam_protocol.pdf
SAEC	Small airway epithelial cells	N	195,662	279	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/SAEC_Stam_protocol.pdf

Cell_line	Description	Fetal?	#DHSs	#SNPs	Pub?	Cell/Tissue Isolation/Culture Protocol
SK-N-SH_RA	Neuroblastoma cell lines differentiated with retinoic acid	N	78,279	99	Y	Stam_protocol.pdf genome.ucsc.edu/ENCODE/protocols/cell/human/Stam_15_protocols.pdf
SK_N_MC	Neuroepithelioma cell line derived from a metastatic supra-orbital human brain tumor	N	154,275	177	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/SK-N-MC_Stam_protocol.pdf
SKMC	Human skeletal muscle cells	Y	208,844	274	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/SkMC_Stam_protocol.pdf
WERI-Rb1	Retinoblastoma cell line	N	190,883	257	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/WERI-Rb-1_Stam_protocol.pdf
WI-38	Embryonic lung fibroblasts immortalized hTERT	Y	164,321	252	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/WI38_Stam_protocol.pdf
WI-38_TAM	Embryonic lung fibroblasts immortalized hTERT_Tamoxifin treated	Y	206,929	358	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/WI38_Stam_protocol.pdf
fAdrenal	Fetal adrenal tissue, 5 samples, ages 7-12 weeks	Y	282,181	480	N	roadmapepigenomics.org/files/protocols/experimental/dnaseI-sensitivity/Nuclei_isolation_DNase_Treatment_Human_tissue_DoucningV4_UW-NREMC.pdf
fBrain	Fetal brain tissue, 12 samples, ages 12-20 weeks	Y	441,136	621	N	roadmapepigenomics.org/files/protocols/experimental/dnaseI-sensitivity/Nuclei_isolation_DNase_Treatment_Human_tissue_DoucningV4_UW-NREMC.pdf
fHeart	Fetal heart tissue, 12 samples, ages 13-21 weeks	Y	393,615	743	N	roadmapepigenomics.org/files/protocols/experimental/dnaseI-sensitivity/Nuclei_isolation_DNase_Treatment_human_tissue-gentleMACS_V5_UW-NREMC.pdf
fIntestine_Lg	Fetal large-intestine tissue, 15 samples, ages 12-16 weeks	Y	439,553	839	N	roadmapepigenomics.org/files/protocols/experimental/dnaseI-sensitivity/Nuclei_isolation_DNase_Treatment_human_tissue-gentleMACS_V5_UW-NREMC.pdf
fIntestine_Sm	Fetal small-intestine tissue, 13 samples, ages 12-16 weeks	Y	360,316	735	N	roadmapepigenomics.org/files/protocols/experimental/dnaseI-sensitivity/Nuclei_isolation_DNase_Treatment_human_tissue-gentleMACS_V5_UW-NREMC.pdf
fKidney	Fetal kidney tissue, 47 samples, ages 12-21 weeks	Y	666,350	1124	N	roadmapepigenomics.org/files/protocols/experimental/dnaseI-sensitivity/Nuclei_isolation_DNase_Treatment_Human_tissue_DoucningV4_UW-NREMC.pdf
fLung	Fetal lung tissue, 34 samples, ages 10-17 weeks	Y	442,491	917	N	roadmapepigenomics.org/files/protocols/experimental/dnaseI-sensitivity/Nuclei_isolation_DNase_Treatment_Human_tissue_DoucningV4_UW-NREMC.pdf
fMuscle	Fetal muscle tissue, 48 samples, ages 12-18 weeks	Y	632,517	1176	N	roadmapepigenomics.org/files/protocols/experimental/dnaseI-sensitivity/Nuclei_isolation_DNase_Treatment_human_tissue-gentleMACS_V5_UW-NREMC.pdf
fPlacenta	Placenta tissue, 4 samples, ages 12-15 weeks	Y	281,754	553	N	roadmapepigenomics.org/files/protocols/experimental/dnaseI-sensitivity/Nuclei_isolation_DNase_Treatment_human_tissue-gentleMACS_V5_UW-NREMC.pdf
fSkin	Fetal fibroblasts, 17 samples, ages 12-14 weeks	Y	392,999	591	N	N/A
fSpinal_cord	Fetal spinal-cord tissue, 3 samples, ages 12-16 weeks	Y	320,476	554	N	roadmapepigenomics.org/files/protocols/experimental/dnaseI-sensitivity/Nuclei_isolation_DNase_Treatment_human_tissue-gentleMACS_V5_UW-NREMC.pdf , but with gentleMACSDissociator Program "B.01 C Tube"
fSpleen	Fetal spleen tissue, age 16 weeks	Y	175,572	334	N	roadmapepigenomics.org/files/protocols/experimental/dnaseI-sensitivity/Nuclei_isolation_DNase_Treatment_Human_tissue_DoucningV4_UW-NREMC.pdf
fStomach	Fetal stomach tissue, 11 samples, ages 13-21 weeks	Y	346,348	658	N	roadmapepigenomics.org/files/protocols/experimental/dnaseI-sensitivity/Nuclei_isolation_DNase_Treatment_human_tissue-gentleMACS_V5_UW-NREMC.pdf
fTestes	Fetal testicle tissue, age 16 weeks	Y	170,843	309	N	roadmapepigenomics.org/files/protocols/experimental/

Cell_line	Description	Fetal?	#DHSs	#SNPs	Pub?	Cell/Tissue Isolation/Culture Protocol
fThymus	Fetal thymus tissue, 10 samples, ages 12-21 weeks	Y	341,548	658	N	dnasel-sensitivity/Nuclei_isolation_DNase_Treatment_Human_tissue_DoucningV4_UW-NREMC.pdf roadmapepigenomics.org/files/protocols/experimental/dnasel-sensitivity/Nuclei_isolation_DNase_Treatment_Human_tissue_DoucningV4_UW-NREMC.pdf
Th1	Human primary T helper 1 cells	N	70,474	141	N	N/A
Th1	Human primary T helper 1 cells	N	73,754	190	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/Stam_15_protocols.pdf
Th17	Human primary T helper 17 cells	N	78,543	130	N	N/A
Th2	Human primary T helper 2 cells	N	111,450	220	N	N/A
Th2	Human primary T helper 2 cells	N	80,196	201	Y	genome.ucsc.edu/ENCODE/protocols/cell/human/Th2_Stam_protocol.pdf
iPS_19_11	Induced pluripotent stem cells	-	204,668	215	N	Yu et al., Cell Stem Cell 8, 326-334 (2011)
iPS_19_7	Induced pluripotent stem cells	-	185,193	199	N	Yu et al., Cell Stem Cell 8, 326-334 (2011)
iPS_4_7	Induced pluripotent stem cells	-	193,671	226	N	Yu et al., Cell Stem Cell 8, 326-334 (2011)
iPS_6_9	Induced pluripotent stem cells	-	191,788	239	N	Yu et al., Cell Stem Cell 8, 326-334 (2011)
vHMEC	Human Mammary Epithelial Cells	N	161,796	272	N	N/A

Table S2. Noncoding SNP-trait associations.

5,654 noncoding GWAS SNP-trait associations, corresponding to 5,134 unique SNPs. Internal-ly_replicated, association was replicated inside the original study in a second population; inde-pendently_replicated, association was replicated in a second population in a separate published study. In_DHS, whether the SNP overlaps a DHS. Fetal_origin, whether the tissue is of fetal origin. Coordinates are hg19, 0-indexed.

NOTE: Due to the size of this file, this table has been uploaded as a separate file.

Table S3. Enrichment of GWAS and control sites for DHSs.

Evaluation of factors that might contribute to enrichment of sites within DHSs. Mean minor allele frequency (MAF) within the CEU population was computed using 1000 Genomes data for all except HapMap. Standard deviation (SD) of the MAF was ~0.14 in all cases; SD was ~1% of the mean for each reported %CG value. Only noncoding sites were surveyed for this table. Note that HapMap SNPs are not distinguished by G+C content. Further, although they are not enriched for introns, within introns, HapMap SNPs are enriched for DHSs.

Sites	% in DHSs	Enrichment for DHSs	Mean MAF	Median distance to nearest TSS (kb)	Mean %CG (± 10 bp)	% in introns	%Intronic sites in DHSs
Random 36mer-mappable sites	41.1%	1.00	NA	71.4	40.16	36.7%	47.5%
1000 Genomes CEU SNPs	42.9%	1.04	0.18	77.7	41.01	35.9%	49.6%
HapMap CEU SNPs	46.5%	1.13	0.22	84.1	40.95	37.7%	52.9%
Affymetrix 500k SNPs	49.7%	1.21	0.23	82.2	40.85	37.5%	55.9%
Unreplicated GWAS SNPs	53.2%	1.29	0.24	57.0	41.66	40.2%	60.3%
Internally replicated GWAS SNPs	59.5%	1.45	0.28	31.4	42.97	45.2%	61.9%
Externally replicated GWAS SNPs	69.8%	1.70	0.28	24.1	43.21	48.1%	71.2%
All GWAS SNPs	57.1%	1.39	0.26	40.7	42.36	43%	62%

Table S4. Cell types used in DHS correlation analysis from Thurman et al.

The 79 cell types in Thurman et al. (10) were assigned to 32 numbered cell-type categories.

Category	Cell types represented
1	WERI_Rb1
2	BE_2_C
3	CACO2, HEPG2, SKNSH
4	HESC, hESCT0
5	A549, HCT116, Hela, PANC1
6	LNCap, MCF7
7	CD56, CD4, hTH1, hTH2
8	GM06990, GM12864, GM12865, GM12878
9	CD34, Jurkat
10	K562, CMK
11	NB4, HL60, CD14, HRGEC
12	HMVEC_LBL, HMVEC_dLyNeo, HMVEC_dBlAd, HMVEC_dBlNeo, HUVEC
13	HMVEC_dLyAd, HMVEC_dNeo
14	NHLF, NHA
15	HAc
16	HAsp
17	HVMF
18	HAEpiC
19	WI_38, AG04450, IMR90
20	SkMC
21	HCFaa
22	HBMEC, HCPEpiC, HIPEpiC, HNPCEpiC
23	HSMM, HSMM_D
24	HCF, HCM, HPAF
25	AG04449, AG09309, AG10803, BJ, HFF
26	NHDF_Ad, HDF_Neo
27	AoAF, HConF, HMF, HPF
28	AG09319, HGF, HPdLF
29	HRCE, HRE, RPTEC
30	HRPEpiC
31	HMEC, NHEK
32	HEEpiC, SAEC

Table S5. New cell types used in DHS correlation analysis

For the correlation analysis, 46 additional cell and tissue types (choosing a representative single time point from each fetal tissue vs. all developmental time points) were condensed to 35 numbered categories.

Category	Cell types represented
1	fAdrenal
2	fBrain
3	fHeart
4	fIntestine_Lg
5	fIntestine_Sm
6	fKidney
7	fLung
8	fMuscle_leg
9	fPlacenta
10	fSkin_fibro_scalp
11	fSpinal_cord
12	fSpleen
13	fStomach
14	fTestes
15	fThymus
16	Skin_Fibroblasts
17	Skin_Keratinocytes
18	Skin_Melanocytes
19	CD20+
20	HAh
21	HFF_MyC
22	HPAEC
23	Mesendoderm
24	NPC
25	NT2_D1
26	PrEC
27	SK_N_MC
28	Trophoblast
29	vHMEC
30	WI_38_TAM
31	HMVEC_dAd, HMVEC_dNeo
32	iPS_19_7, iPS_19_11, iPS_4_7
33	iPS_6_9, H1_P18, H9_P42
34	CD3+_CordBlood, CD19+, CD20+
35	CD8+, CD3+, hTH1+, hTH17+

Table S6. Genes correlated with distal DHSs harboring GWAS SNPs (Part I)

347 trait-SNP associations (296 unique SNPs) overlapping predicted long-distance interactions established by correlation of chromatin accessibility (r) in Thurman et al. (10). LD measures the mean extent of LD between the correlated DHSs (r^2); NA means filtered 1000 Genomes SNPs with MAF \geq 5% in the CEU population were not found within 2 kb of both DHSs. Cor_gene_name represents the most-correlated gene; Dist, distance to gene in kb. "Adjacent?", whether the highest-correlated gene is an adjacent gene.

SNP	Disease or trait	Trait Category	r	LD	Cor_gene_name	Dist	Adjacent?
rs10202497	Aging_traits-age_free_from_disease	Aging	0.75	0.05	COL6A3	52	Y
rs10781380	Hippocampal_atrophy	Aging	0.93	0.01	FOXB2	226	N
rs11006923	Alzheimers_disease	Aging	0.70	0.06	RP11-351M16.2	128	N
rs1172822	Menopause	Aging	0.95	0.01	NLRP2	-347	N
rs12203592	Progressive_supranuclear_palsy	Aging	0.85	0.01	RP3-416J7.3	-241	N
rs1532278	Alzheimers_disease_late_onset	Aging	0.81	0.01	RP11-16P20.2	89	N
rs1561570	Pagets_disease	Aging	0.71	0.03	RP11-730A19.2	-53	N
rs1564282	Parkinsons_disease	Aging	0.88	0.05	CPLX1	-54	N
rs1569476	Alzheimers_Total_ventricular_volume	Aging	0.98	0.03	SCYL3	250	N
rs157580	Alzheimers-AB1-42	Aging	0.85	0.00	CEACAM19	-221	N
rs157580	Alzheimers_disease	Aging	0.85	0.00	CEACAM19	-221	N
rs16938437	Menarche	Aging	0.89	0.29	PHF21A	-71	Y
rs1695739	Longevity	Aging	0.78	0.01	DDX25	-405	N
rs1934951	Osteonecrosis_of_the_jaw	Aging	0.88	0.16	RP11-310E22.4	192	N
rs2104362	Amyotrophic_lateral_sclerosis-age_of_onset	Aging	1.00	0.02	SYNGAP1	-412	N
rs2121433	Alzheimers-t-tau	Aging	0.76	0.01	AC105402.1	65	Y
rs2244621	Longevity	Aging	0.92	0.01	RASGRP2	484	N
rs2687729	Menarche	Aging	0.73	0.01	DNAJB8	291	N
rs2899472	Alzheimers-AB1-42	Aging	0.74	0.03	GLDN	153	N
rs3825776	Amyotrophic_lateral_sclerosis	Aging	0.71	0.02	AQP9	-316	N
rs4955755	Menopause	Aging	0.87	0.01	CLDN11	-339	N
rs5937496	Amyotrophic_lateral_sclerosis	Aging	0.86	N/A	SAR1AP4	-288	N
rs6031882	Hippocampal_atrophy	Aging	0.72	0.01	BLCAP	348	N
rs6602175	Alzheimers-Whole-brain_volume	Aging	0.77	0.02	TRDMT1	101	N
rs6701713	Alzheimers_disease_late_onset	Aging	0.85	0.00	PLXNA2	428	N
rs9652490	Essential_tremor	Aging	0.77	0.01	SH2D7	421	N
rs9871760	Alzheimers-Whole-brain_volume	Aging	0.82	0.12	RP11-305F5.2	-52	Y
rs10036748	Systemic_lupus_erythematosus	Autoimmune	0.78	0.01	ANXA6	63	N
rs10737562	Systemic_lupus_erythematosus	Autoimmune	0.83	0.65	RP11-398M15.1	21	Y
rs1077667	Multiple_sclerosis	Autoimmune	0.80	0.01	GTF2F1	-277	N
rs10931468	Primary_biliary_cirrhosis	Autoimmune	0.81	0.03	TMEM194B	-147	N
rs11154801	Multiple_sclerosis	Autoimmune	0.90	0.15	AH1	-95	Y
rs11616188	Ankylosing_spondylitis	Autoimmune	0.91	0.01	NCAPD2	122	N
rs11742570	Crohns_disease	Autoimmune	0.77	0.03	PTGER4	271	Y
rs12212193	Multiple_sclerosis	Autoimmune	0.73	0.01	GJA10	-394	N
rs12580100	Psoriasis	Autoimmune	0.73	0.05	SLC39A5	185	N
rs1295686	Asthma	Autoimmune	0.91	0.24	AC004041.2	4	Y
rs1335532	Multiple_sclerosis	Autoimmune	0.92	0.01	CD101	443	N
rs1551398	Crohns_disease	Autoimmune	1.00	0.00	TRIB1	-95	N
rs17035378	Celiac_disease	Autoimmune	0.87	0.02	ARHGAP25	363	N
rs17582416	Crohns_disease	Autoimmune	0.71	0.51	RP11-324I22.2	232	N
rs17716942	Psoriasis	Autoimmune	0.71	0.01	AC007740.1	365	N
rs1790100	Multiple_sclerosis	Autoimmune	0.98	0.01	RP11-324E6.4	-422	N
rs1800871	Behcets_disease	Autoimmune	0.88	0.01	PFKFB2	297	N
rs2056626	Systemic_sclerosis	Autoimmune	0.82	0.01	ILDR2	-476	N
rs2075726	Ankylosing_spondylitis	Autoimmune	0.88	0.01	LL22NC01-81G9.3	50	N
rs2104286	Multiple_sclerosis	Autoimmune	0.70	0.11	IL2RA	-29	Y
rs2187668	Systemic_lupus_erythematosus	Autoimmune	0.88	0.01	BRD2	335	N
rs2205960	Systemic_lupus_erythematosus	Autoimmune	0.91	0.01	SLC9A11	379	N
rs2233287	Systemic_sclerosis	Autoimmune	0.86	0.02	ANXA6	68	N

SNP	Disease or trait	Trait Category	r	LD	Cor_gene_name	Dist	Adjacent?
rs2273017	Graves_disease	Autoimmune	0.72	0.03	CFB	-424	N
rs2431697	Systemic_lupus_erythematosus	Autoimmune	0.85	0.01	PWWP2A	-334	N
rs2546890	Multiple_sclerosis	Autoimmune	0.87	0.02	EBF1	-313	N
rs2546890	Psoriasis	Autoimmune	0.87	0.02	EBF1	-313	N
rs2618476	Systemic_lupus_erythematosus	Autoimmune	0.93	0.11	BLK	50	Y
rs2734583	Stevens-Johnson_syndrome_and_necrolysis	Autoimmune	0.80	0.04	WASF5P	-249	N
rs2836878	Inflammatory_bowel_disease	Autoimmune	0.80	0.03	LCA5L	352	N
rs2836878	Ulcerative_colitis	Autoimmune	0.80	0.03	LCA5L	352	N
rs3024505	Crohns_disease	Autoimmune	0.79	0.33	IL10	6	Y
rs3024505	Ulcerative_colitis	Autoimmune	0.79	0.33	IL10	6	Y
rs3129763	Systemic_sclerosis	Autoimmune	0.98	N/A	HLA-DRA	-183	N
rs3821236	Systemic_sclerosis	Autoimmune	0.74	0.02	STAT4	113	Y
rs3821236	Systemic_lupus_erythematosus	Autoimmune	0.74	0.02	STAT4	113	Y
rs4075958	Multiple_sclerosis	Autoimmune	0.90	0.02	GRK6	74	N
rs4129267	Asthma	Autoimmune	0.88	0.39	IL6R	-19	Y
rs4349859	Ankylosing_spondylitis	Autoimmune	0.92	0.02	POU5F1	-229	N
rs4409764	Crohns_disease	Autoimmune	0.90	0.02	DNMBP	390	N
rs4639966	Systemic_lupus_erythematosus	Autoimmune	0.99	N/A	NLRX1	473	N
rs4781011	Ulcerative_colitis	Autoimmune	0.75	0.01	AC007014.1	144	N
rs4845783	Asthma	Autoimmune	0.81	0.04	LCE3A	103	N
rs485499	Primary_biliary_cirrhosis	Autoimmune	0.71	0.01	SMC4	392	N
rs5754217	Systemic_lupus_erythematosus	Autoimmune	0.86	N/A	PI4KAP2	-73	N
rs6074022	Multiple_sclerosis	Autoimmune	0.96	0.00	SLC35C2	252	N
rs610604	Psoriasis	Autoimmune	0.84	0.29	TNFAIP3	-11	Y
rs6806528	Celiac_disease	Autoimmune	0.89	0.18	FRMD4B	-3	Y
rs6859219	Rheumatoid_arthritis	Autoimmune	0.75	0.09	RPL17P22	-13	Y
rs6941421	Multiple_sclerosis	Autoimmune	0.96	0.22	RP1-190J20.2	18	N
rs734999	Ulcerative_colitis	Autoimmune	0.84	0.00	C1orf86	-369	N
rs7579944	Rheumatoid_arthritis_celiac_disease	Autoimmune	0.84	0.01	AC016907.2	-146	N
rs794185	Multiple_sclerosis-Brain_Glutamate_Concentrations	Autoimmune	0.75	0.01	ITPR1	419	N
rs806321	Multiple_sclerosis	Autoimmune	1.00	0.10	DLEU1	43	Y
rs881375	Rheumatoid_arthritis	Autoimmune	0.97	0.07	RP11-27I1.2	-44	N
rs924080	Behcets_disease	Autoimmune	0.87	0.01	IL12RB2	13	Y
rs943072	Ulcerative_colitis	Autoimmune	0.92	0.01	SLC35B2	429	N
rs987870	Asthma	Autoimmune	0.92	0.06	MYL8P	261	N
rs988739	Systemic_lupus_erythematosus	Autoimmune	0.97	0.03	STX4	-265	N
rs10510102	Breast_cancer	Cancer	0.92	0.01	RPS15AP5	-152	N
rs11892031	Bladder_cancer	Cancer	0.92	0.03	AC019221.4	-302	N
rs12653946	Prostate_cancer	Cancer	0.92	0.04	RP11-259Q02.3	72	N
rs13397985	Chronic_lymphocytic_leukemia	Cancer	0.97	0.05	AC009950.1	-23	N
rs1432295	Hodgkins_lymphoma	Cancer	0.96	N/A	AC007381.3	-486	N
rs16886165	Breast_cancer	Cancer	0.88	0.23	MAP3K1	158	N
rs2157719	Glioma	Cancer	0.97	0.00	RP11-344A7.1	-398	N
rs2456449	Chronic_lymphocytic_leukemia	Cancer	0.87	0.05	RP11-255B23.2	-98	N
rs28421666	Nasopharyngeal_carcinoma	Cancer	0.85	0.02	BRD2	348	N
rs339331	Prostate_cancer	Cancer	0.97	0.03	FAM26D	-335	N
rs402710	Lung_cancer	Cancer	0.88	0.39	CLPTM1L	18	Y
rs4132601	Acute_lymphoblastic_leukemia_childhood	Cancer	0.75	0.02	AC020743.3	-226	N
rs4487645	Multiple_myeloma	Cancer	0.96	0.01	DNAH11	-344	Y
rs4975616	Lung_cancer	Cancer	0.96	0.35	CLPTM1L	16	Y
rs4980785	Renal_cell_carcinoma	Cancer	0.77	0.01	RP11-300I6.5	322	N
rs498872	Glioma	Cancer	0.71	0.02	SLC37A4	423	N
rs674313	Chronic_lymphocytic_leukemia	Cancer	0.92	0.05	PSMB8	232	N
rs7579899	Renal_cell_carcinoma	Cancer	0.72	0.02	RP11-417F21.2	188	N
rs961253	Colorectal_cancer	Cancer	0.96	0.01	RP5-859D4.3	312	N
rs10765792	Sudden_cardiac_arrest	Cardiovascular	0.90	0.01	FAM76B	-350	N
rs11710077	QRS_duration	Cardiovascular	0.96	0.02	SCN10A	181	N
rs12046278	Systolic_blood_pressure	Cardiovascular	0.87	0.01	MASP2	308	N
rs12576239	QT_interval	Cardiovascular	0.98	N/A	ASCL2	-210	N
rs1378942	Diastolic_blood_pressure	Cardiovascular	0.84	0.09	SEMA7A	-351	N
rs1378942	Systolic_blood_pressure	Cardiovascular	0.84	0.09	SEMA7A	-351	N
rs1378942	Blood_pressure	Cardiovascular	0.84	0.09	SEMA7A	-351	N
rs16857031	QT_interval	Cardiovascular	0.92	0.01	OLFML2B	-157	N
rs16933812	Blood_pressure	Cardiovascular	0.82	0.02	RP11-397D12.7	465	N
rs17259784	Cardiac_hypertrophy	Cardiovascular	0.71	0.04	RP11-565N2.2	49	N

SNP	Disease or trait	Trait Category	r	LD	Cor_gene_name	Dist	Adjacent?
rs1746048	Coronary_heart_disease	Cardiovascular	0.98	0.01	RP11-73D4.1	327	N
rs1746048	Myocardial_infarction	Cardiovascular	0.98	0.01	RP11-73D4.1	327	N
rs17672135	Coronary_heart_disease	Cardiovascular	0.76	0.06	FMN2	-47	Y
rs17691394	Carotid_atherosclerosis_in_HIV_infection	Cardiovascular	0.92	0.00	GRM8	430	N
rs190759	Sudden_cardiac_arrest	Cardiovascular	0.84	0.01	TFAP2B	-198	N
rs2074238	QT_interval	Cardiovascular	0.73	0.01	AC013791.2	307	Y
rs4638289	Atherosclerosis	Cardiovascular	0.98	0.01	TSG101	222	N
rs4687718	QRS_duration	Cardiovascular	0.89	N/A	TMEM110-MUSTN1	-405	N
rs54211	Sudden_cardiac_arrest	Cardiovascular	0.73	0.01	CTA-150C2.16	-293	N
rs6801957	QRS_duration	Cardiovascular	0.85	0.04	SCN10A	72	Y
rs7808424	Coronary_heart_disease	Cardiovascular	0.86	0.37	AC003045.1	15	N
rs789852	QT_interval	Cardiovascular	0.93	0.01	ATP13A3	-146	N
rs8049607	QT_interval	Cardiovascular	0.70	0.01	PRM1	-314	N
rs880315	Diastolic_blood_pressure	Cardiovascular	0.97	N/A	RP11-340B24.3	157	N
rs880315	Systolic_blood_pressure	Cardiovascular	0.97	N/A	RP11-340B24.3	157	N
rs9298506	Intracranial_aneurysm	Cardiovascular	0.95	0.35	RP11-53M11.3	28	Y
rs944260	Sudden_cardiac_arrest	Cardiovascular	0.77	0.01	RP11-429E11.3	52	Y
rs9470361	QRS_duration	Cardiovascular	0.84	0.01	RP1-90K10.3	272	N
rs9581094	Sudden_cardiac_arrest	Cardiovascular	0.73	0.23	PARP4	4	Y
rs964184	Coronary_heart_disease	Cardiovascular	0.90	0.04	SIK3	96	N
rs11867934	Diabetic_retinopathy	Diabetes	0.96	N/A	FLCN	195	N
rs17696736	Type_1_diabetes	Diabetes	0.75	0.08	ACAD10	-343	N
rs2237897	Type_2_diabetes	Diabetes	0.72	0.01	OSBPL5	253	N
rs3007729	Diabetic_retinopathy	Diabetes	0.82	0.02	IGSF21	-95	N
rs3024505	Type_1_diabetes_autoantibodies	Diabetes	0.79	0.33	IL10	6	Y
rs3024505	Type_1_diabetes	Diabetes	0.79	0.33	IL10	6	Y
rs5753037	Type_1_diabetes	Diabetes	0.77	0.26	HORMAD2	-63	N
rs7111341	Type_1_diabetes	Diabetes	0.95	0.02	IGF2	-43	N
rs7171171	Type_1_diabetes_autoantibodies	Diabetes	0.74	0.04	C15orf53	82	Y
rs10202231	Response_to_antipsychotic_therapy_perphenazine-triglycerides	Drug_metabolism	0.99	0.01	RP11-416L21.1	438	N
rs1061235	Response_to_carbamapexine	Drug_metabolism	0.83	0.14	HLA-A	-3	N
rs10950821	Response_to_statin_therapy-acylcarnitine	Drug_metabolism	0.84	0.01	MACC1	-390	N
rs12147450	Response_to_antipsychotic_therapy_extrapyramidal_side_effects	Drug_metabolism	0.77	0.01	CCNB1IP1	-160	N
rs1535	Response_to_statin_therapy-braces	Drug_metabolism	0.92	0.01	C11orf66	-342	N
rs2163287	Response_to_antidepressants-bupropion	Drug_metabolism	0.97	N/A	SERAC1	499	N
rs2830840	Response_to_citalopram_treatment	Drug_metabolism	0.71	0.01	AP001601.2	-404	N
rs286913	Response_to_antipsychotic_therapy-FEV1/FVC	Drug_metabolism	0.96	0.01	ELF5	-120	N
rs2954038	Response_to_statin_therapy-Triglyceride_sum	Drug_metabolism	0.99	0.02	TRIB1	-62	N
rs3753242	Olanzapine_Schizophrenia_neurocognition	Drug_metabolism	0.91	0.45	PRKCZ	-3	Y
rs3795578	Acetaminophen_hepatotoxicity	Drug_metabolism	1.00	0.01	RP11-203F10.6	204	N
rs9658108	Response_to_antipsychotic_therapy_clozapine-glucose	Drug_metabolism	0.73	0.33	ZNF76	-105	N
rs1034566	Platelet_count	Hematological_param	0.99	0.23	ARVCF	-6	Y
rs10489087	Red_blood_cell_count	Hematological_param	0.94	0.01	RP11-341G5.1	23	Y
rs11628318	Platelet_count	Hematological_param	0.89	0.02	RAGE	-335	Y
rs12566888	Platelet_aggregation-ADP	Hematological_param	0.87	0.00	IQGAP3	-360	N
rs12566888	Platelet_aggregation-epinephrine	Hematological_param	0.87	0.00	IQGAP3	-360	N
rs12718597	Mean_corpuscular_volume	Hematological_param	0.72	0.01	AC020743.3	-184	N
rs1354034	Mean_platelet_volume	Hematological_param	0.92	N/A	CCDC66	-199	N
rs1354034	Platelet_count	Hematological_param	0.92	N/A	CCDC66	-199	N
rs1408272	Mean_corpuscular_hemoglobin	Hematological_param	0.70	0.06	TRIM38	124	N
rs1558324	Mean_platelet_volume	Hematological_param	0.87	0.13	VWF	-55	N
rs2336384	Platelet_count	Hematological_param	0.80	0.05	MIIP	34	N
rs385893	Platelet_count	Hematological_param	0.97	0.01	JAK2	221	N
rs3859192	WBC_count	Hematological_param	0.85	0.02	RARA	336	N
rs4148441	Platelet_count	Hematological_param	0.77	0.01	ABCC4	-188	Y
rs4660456	Platelet_count	Hematological_param	0.93	0.01	COL9A2	-456	N
rs4812048	Mean_platelet_volume	Hematological_param	0.98	0.00	EDN3	288	N
rs4895441	Mean_corpuscular_volume	Hematological_param	0.86	0.08	RP1-32B1.4	155	N
rs4895441	WBC_count	Hematological_param	0.86	0.08	RP1-32B1.4	155	N
rs6108011	Red_blood_cell_count	Hematological_param	0.73	0.02	RP5-83E8.1	264	Y
rs643381	Mean_corpuscular_volume	Hematological_param	0.81	0.00	RP11-15H7.1	100	Y
rs7775698	Mean_corpuscular_hemoglobin	Hematological_param	0.80	0.05	RP1-32B1.4	162	N
rs7775698	Mean_corpuscular_volume	Hematological_param	0.80	0.05	RP1-32B1.4	162	N

SNP	Disease or trait	Trait Category	r	LD	Cor_gene_name	Dist	Adjacent?
rs7775698	Red_blood_cell_traits	Hematological_param	0.80	0.05	RP1-32B1.4	162	N
rs7961894	Mean_platelet_volume	Hematological_param	0.96	0.01	CLIP1	395	N
rs7961894	Platelet_count	Hematological_param	0.96	0.01	CLIP1	395	N
rs8176746	Mean_corpuscular_hemoglobin	Hematological_param	0.88	0.05	C9orf7	193	N
rs9349205	Mean_corpuscular_hemoglobin	Hematological_param	0.79	0.03	TFEB	-221	N
rs9349205	Mean_corpuscular_volume	Hematological_param	0.79	0.03	TFEB	-221	N
rs9483788	Hematocrit	Hematological_param	0.81	N/A	HBS1L	-130	Y
rs10516526	FEV1	Kidney_lung_liver	0.83	0.08	NPNT	143	N
rs1529672	FEV1/FVC	Kidney_lung_liver	0.81	0.01	TOP2B	140	Y
rs1883414	IgA_nephropathy	Kidney_lung_liver	0.89	N/A	RXRB	82	N
rs2187668	idiopathic_membranous_nephropathy	Kidney_lung_liver	0.88	0.01	BRD2	335	N
rs2216228	NAFLD_histology	Kidney_lung_liver	0.90	0.06	RP11-268P4.2	383	N
rs2284746	FEV1/FVC	Kidney_lung_liver	0.91	0.43	MFAP2	0	Y
rs4129267	FEF	Kidney_lung_liver	0.88	0.39	IL6R	-19	Y
rs643608	NAFLD_histology	Kidney_lung_liver	0.83	0.01	CBS	-279	N
rs7632299	NAFLD_histology	Kidney_lung_liver	0.91	0.01	SLC9A9	360	N
rs10194115	Erectile_dysfunction_and_prostate_cancer_treatment	Miscellaneous	0.99	0.01	C2orf61	139	N
rs12045440	Goiter	Miscellaneous	0.95	N/A	UBR4	-270	N
rs12045440	Thyroid_volume	Miscellaneous	0.95	N/A	UBR4	-270	N
rs13208776	Vitiligo	Miscellaneous	0.89	0.01	FRMD1	-469	N
rs2280543	Uterine_fibroids	Miscellaneous	0.88	0.01	RN1	299	N
rs2553268	Exercise_treadmill_test_traits	Miscellaneous	0.74	0.01	CTD-2373N4.1	-437	N
rs3796619	Recombination_rate_males	Miscellaneous	0.95	0.01	GAK	-246	N
rs6049375	Erectile_dysfunction_and_prostate_cancer_treatment	Miscellaneous	0.79	0.04	GAPDHP53	367	N
rs6847149	Exercise_treadmill_test_traits	Miscellaneous	0.90	0.03	AC004067.4	-205	N
rs735860	Glaucoma	Miscellaneous	0.86	0.01	RP1-214M20.3	-312	N
rs738322	Cutaneous_nevi	Miscellaneous	0.78	0.01	RP1-199H16.5	483	N
rs7567389	Self-rated_health	Miscellaneous	0.82	0.16	MAP3K2	119	N
rs10893366	Alcohol_dependence	Neurological_behavioral	0.71	0.02	E124	271	N
rs1107592	Bipolar_disorder_and_schizophrenia	Neurological_behavioral	0.83	0.04	MAD1L1	215	Y
rs12290811	Bipolar_disorder	Neurological_behavioral	0.98	0.01	ODZ4	-102	Y
rs12807809	Schizophrenia	Neurological_behavioral	0.99	0.01	SLC37A2	327	N
rs1412115	Schizophrenia	Neurological_behavioral	0.80	0.07	RP11-490O24.1	64	Y
rs1449984	Major_depressive_disorder	Neurological_behavioral	0.95	0.02	AC016768.1	-158	Y
rs1550976	Asperger_disorder	Neurological_behavioral	0.93	0.00	AP002856.5	-197	N
rs16973500	ADHD	Neurological_behavioral	0.91	0.08	PMFBP1	240	N
rs17069122	Bipolar_disorder_and_schizophrenia	Neurological_behavioral	0.72	0.14	RP1-111B22.2	4	Y
rs1879248	Schizophrenia	Neurological_behavioral	0.81	0.21	FXR1	120	Y
rs2002030	Immediate_Story_Recall	Neurological_behavioral	0.81	0.04	BLK	75	N
rs2021722	Schizophrenia	Neurological_behavioral	0.70	0.11	KIAA1949	482	N
rs2070615	Bipolar_disorder	Neurological_behavioral	0.95	0.02	RPS10P20	-337	N
rs2268983	Smoking_behavior	Neurological_behavioral	0.76	0.03	EXD2	285	N
rs2349775	Neuroticism	Neurological_behavioral	0.90	0.00	ICA1	-415	N
rs4307059	Autism	Neurological_behavioral	0.85	0.19	MSNP1	-60	Y
rs4380451	Bipolar_disorder	Neurological_behavioral	0.86	0.00	OSBPL10	-368	N
rs493187	Bipolar_disorder_and_schizophrenia	Neurological_behavioral	0.91	0.06	RP11-15J23.1	-327	N
rs6716455	Alcohol_dependence	Neurological_behavioral	0.83	0.10	AC113610.1	-10	Y
rs6716455	Alcohol_use_disorder	Neurological_behavioral	0.83	0.10	AC113610.1	-10	Y
rs6782029	Anorexia_nervosa	Neurological_behavioral	0.94	0.40	VGLL4	0	Y
rs6952808	Bipolar_disorder_and_schizophrenia	Neurological_behavioral	0.89	0.16	MAD1L1	-15	N
rs6968385	ADHD	Neurological_behavioral	0.93	0.10	AC003088.1	127	Y
rs702543	Neuroticism	Neurological_behavioral	0.82	0.00	PDE4D	-330	N
rs7045881	Schizophrenia	Neurological_behavioral	0.88	0.01	NCRNA00032	354	N
rs7178909	Common_traits_optimism	Neurological_behavioral	0.73	0.01	IDH2	198	N
rs7520258	Working_memory	Neurological_behavioral	0.92	0.01	LGALS8	391	N
rs7578035	Bipolar_disorder	Neurological_behavioral	0.95	0.08	YWHAQP5	-73	N
rs7581919	Conduct_disorder_case_status	Neurological_behavioral	0.99	0.01	RP11-120J4.1	345	N
rs7992643	ADHD	Neurological_behavioral	0.97	0.05	CLYBL	-32	Y
rs806276	ADHD	Neurological_behavioral	0.76	0.01	BACH2	-489	Y
rs933688	Smoking_behavior	Neurological_behavioral	0.95	0.21	RP11-414H23.2	-245	N
rs9810857	ADHD	Neurological_behavioral	0.80	0.01	RP11-372E1.4	-339	N
rs9845475	ADHD	Neurological_behavioral	0.76	0.10	CNOT10	-31	N
rs1451375	Malaria	Parasitic_bacterial_disease	0.84	0.11	GRB10	52	N
rs10514345	Hip_geometry	Quantitative_traits	0.91	0.03	RP11-414H23.2	94	Y

SNP	Disease or trait	Trait Category	r	LD	Cor_gene_name	Dist	Adjacent?
rs11989122	Height	Quantitative_traits	0.91	0.01	AC023590.1	475	N
rs12203592	Freckling	Quantitative_traits	0.85	0.01	RP3-416J7.3	-241	N
rs12203592	Hair_color-Black_vs_blonde_hair_color	Quantitative_traits	0.85	0.01	RP3-416J7.3	-241	N
rs12203592	Hair_color-Black_vs_red_hair_color	Quantitative_traits	0.85	0.01	RP3-416J7.3	-241	N
rs12203592	Hair_color	Quantitative_traits	0.85	0.01	RP3-416J7.3	-241	N
rs1635852	Height	Quantitative_traits	0.76	0.00	CREB5	285	N
rs2054989	Hip_geometry	Quantitative_traits	0.98	0.25	C3orf63	133	N
rs2282978	Height	Quantitative_traits	0.76	0.02	KRIT1	-392	N
rs2284746	Height	Quantitative_traits	0.91	0.43	MFAP2	0	Y
rs2336725	Height	Quantitative_traits	0.95	0.01	PRKCD	71	N
rs2523178	Height	Quantitative_traits	0.87	0.04	DOT1L	-111	Y
rs2730245	Height	Quantitative_traits	0.84	0.10	NCAPG2	-227	N
rs291671	Hair_color-red_hair	Quantitative_traits	0.83	0.02	RP4-553F4.6	430	N
rs3782089	Height	Quantitative_traits	0.88	0.06	FIBP	317	N
rs3791950	Height	Quantitative_traits	0.72	0.01	PNKD	469	N
rs4072910	Height	Quantitative_traits	0.91	0.01	PRAM1	-87	N
rs4282339	Height	Quantitative_traits	0.98	0.18	SLIT3	15	Y
rs4823006	Waist-hip_ratio	Quantitative_traits	0.81	0.01	AP1B1	315	N
rs4932217	Height	Quantitative_traits	0.83	0.23	POLG	-40	Y
rs619865	Freckling	Quantitative_traits	0.72	0.01	RBM39	457	N
rs6784615	Waist-hip_ratio	Quantitative_traits	0.73	0.22	BAP1	-64	N
rs6899976	Height	Quantitative_traits	0.71	0.01	RP1-69D17.4	-490	N
rs7007970	Height	Quantitative_traits	0.88	0.00	RP11-775B15.3	-152	N
rs7121446	Waist_circumference	Quantitative_traits	0.80	0.02	RP11-166D19.1	18	Y
rs7349332	Hair_curl	Quantitative_traits	0.72	0.18	AC097468.6	63	N
rs7349332	Hair_morphology	Quantitative_traits	0.72	0.18	AC097468.6	63	N
rs735854	Optic_disc_size_rim	Quantitative_traits	0.72	0.01	APOL3	-117	N
rs7466269	Height	Quantitative_traits	0.99	0.13	RP11-57C19.2	57	N
rs798497	Height	Quantitative_traits	0.95	0.00	EIF3B	-379	N
rs941873	Height	Quantitative_traits	0.79	0.43	RP11-342M3.5	2	Y
rs946053	Height	Quantitative_traits	0.95	0.01	AMBP	-210	N
rs228769	Bone_mineral_density-hip	Radiographic_parameter	0.71	0.07	MPP2	-215	Y
rs228769	Bone_mineral_density-spine	Radiographic_parameter	0.71	0.07	MPP2	-215	Y
rs4870044	Bone_mineral_density-hip	Radiographic_parameter	0.81	0.04	RP11-351K16.4	-164	N
rs4870044	Bone_mineral_density-spine	Radiographic_parameter	0.81	0.04	RP11-351K16.4	-164	N
rs1039302	C-reactive_protein	Serum_metabolites	0.94	0.01	RNF10	-265	N
rs10889353	Cholesterol	Serum_metabolites	0.96	0.02	RP5-1155K23.1	-463	N
rs10889353	LDL_cholesterol	Serum_metabolites	0.96	0.02	RP5-1155K23.1	-463	N
rs10889353	Triglycerides	Serum_metabolites	0.96	0.02	RP5-1155K23.1	-463	N
rs11597390	Alanine_aminotransferase	Serum_metabolites	0.86	N/A	ENTPD7	-442	N
rs11708067	Fasting_plasma_glucose	Serum_metabolites	0.98	0.03	PDIA5	-223	N
rs11708067	Insulin_resistance	Serum_metabolites	0.98	0.03	PDIA5	-223	N
rs11761528	Serum_dehydroepiandrosterone	Serum_metabolites	0.91	0.31	ARPC1B	-147	N
rs12239046	C-reactive_protein	Serum_metabolites	0.99	0.02	NLRP3	-20	Y
rs12239436	HDL_cholesterol	Serum_metabolites	0.70	0.02	RP11-101C11.1	-59	Y
rs12740374	LDL_cholesterol	Serum_metabolites	0.75	0.01	AMPD2	354	N
rs13022873	Triglycerides_waist_circumference	Serum_metabolites	0.72	0.16	IFT172	-131	N
rs13195786	Serum_calcium	Serum_metabolites	0.85	0.02	TFAP2A	256	N
rs1335645	Gamma_glutamyl_transferase	Serum_metabolites	0.74	N/A	DENND2D	59	N
rs1408272	Transferrin_receptor	Serum_metabolites	0.70	0.06	TRIM38	124	N
rs1535	HDL_cholesterol	Serum_metabolites	0.92	0.01	C11orf66	-342	N
rs1535	Serum_polyunsaturated_fatty_acids	Serum_metabolites	0.92	0.01	C11orf66	-342	N
rs157580	HDL_cholesterol	Serum_metabolites	0.85	0.00	CEACAM19	-221	N
rs157580	LDL_cholesterol	Serum_metabolites	0.85	0.00	CEACAM19	-221	N
rs1594468	Bilirubin	Serum_metabolites	0.74	0.03	ELMOD2	58	N
rs17319721	Creatinine	Serum_metabolites	0.77	N/A	CXCL11	-405	N
rs174536	Serum_polyunsaturated_fatty_acids	Serum_metabolites	0.88	0.00	DAK	-451	N
rs174546	HDL_cholesterol	Serum_metabolites	0.86	0.01	DDB1	-460	N
rs174546	LDL_cholesterol	Serum_metabolites	0.86	0.01	DDB1	-460	N
rs174574	Serum_polyunsaturated_fatty_acids	Serum_metabolites	0.96	0.01	C11orf66	-344	N
rs1967017	Serum_urate	Serum_metabolites	0.76	0.03	RP11-458D21.2	-348	N
rs2052550	Ferritin	Serum_metabolites	0.86	0.08	DMGDH	54	N
rs2066219	Two_hour_glucose_challenge	Serum_metabolites	0.75	0.05	RPL12P34	-311	Y
rs2078267	Gout	Serum_metabolites	0.83	0.00	ARL2	450	N
rs2078267	Serum_urate	Serum_metabolites	0.83	0.00	ARL2	450	N
rs2153960	IGF1	Serum_metabolites	0.97	0.02	NR2E1	-491	N
rs2235302	P-selectin	Serum_metabolites	0.93	0.07	SELL	94	N

SNP	Disease or trait	Trait Category	r	LD	Cor_gene_name	Dist	Adjacent?
rs2650000	C-reactive_protein	Serum_metabolites	0.83	0.01	KDM2B	491	N
rs2650000	LDL_cholesterol	Serum_metabolites	0.83	0.01	KDM2B	491	N
rs2836878	C-reactive_protein	Serum_metabolites	0.80	0.03	LCA5L	352	N
rs2877716	Two-hour_glucose_challenge	Serum_metabolites	0.74	N/A	ADCY5	-56	Y
rs3093030	ICAM1	Serum_metabolites	0.94	0.08	ZGLP1	19	Y
rs3729639	HDL_cholesterol	Serum_metabolites	0.71	N/A	TRADD	-34	N
rs4129267	C-reactive_protein	Serum_metabolites	0.88	0.39	IL6R	-19	Y
rs4129267	IL6R	Serum_metabolites	0.88	0.39	IL6R	-19	Y
rs4273077	Protein-total	Serum_metabolites	0.75	0.01	AC055811.5	294	N
rs4516970	Ferritin	Serum_metabolites	0.76	0.01	RP1-249F5.3	182	N
rs4607517	Insulin_resistance	Serum_metabolites	0.85	0.01	POLD2	-74	N
rs4607517	Fasting_plasma_glucose	Serum_metabolites	0.85	0.01	POLD2	-74	N
rs4686760	Van_Wildebrand_factor_antibodies	Serum_metabolites	0.87	0.02	VPS8	122	Y
rs4737009	HbA1C	Serum_metabolites	0.94	0.03	AGPAT6	-197	N
rs4820599	Gamma_glutamyl_transferase	Serum_metabolites	0.80	0.01	C22orf13	-44	N
rs4963452	Serum_polyunsaturated_fatty_acids	Serum_metabolites	0.80	0.03	SCGB2A2	222	N
rs507666	ICAM1	Serum_metabolites	0.83	0.08	C9orf7	178	N
rs6442522	Serum_urate	Serum_metabolites	0.73	0.06	NR2C2	-451	N
rs6734238	C-reactive_protein	Serum_metabolites	1.00	0.18	IL1RN	44	Y
rs6984305	Alkaline_phosphatase	Serum_metabolites	0.85	0.02	RP11-115J16.2	118	N
rs7117404	Fibrin-D-dimer_levels	Serum_metabolites	0.79	0.01	ATG13	-446	N
rs7120118	HDL_cholesterol	Serum_metabolites	0.79	0.01	CELF1	225	N
rs7569328	LDL_cholesterol	Serum_metabolites	0.80	0.01	HS1BP3	-262	N
rs7778619	CD40_ligand	Serum_metabolites	0.82	0.01	AC060834.2	-385	N
rs8109578	Thyroid_stimulating_hormone	Serum_metabolites	0.92	0.01	PPAN	5	Y
rs911119	Cystatin_C	Serum_metabolites	0.75	0.01	RP4-737E23.4	-499	N
rs964184	Alpha-tocopherol	Serum_metabolites	0.90	0.04	SIK3	96	N
rs964184	HDL_cholesterol	Serum_metabolites	0.90	0.04	SIK3	96	N
rs964184	Hypertriglyceridemia	Serum_metabolites	0.90	0.04	SIK3	96	N
rs964184	Lipoprotein-associated_phospholipase_A2_activity_and_mass	Serum_metabolites	0.90	0.04	SIK3	96	N
rs964184	Triglycerides	Serum_metabolites	0.90	0.04	SIK3	96	N
rs9939224	HDL_cholesterol_fasting_glucose	Serum_metabolites	0.75	0.18	CETP	-7	Y
rs9992101	Creatinine	Serum_metabolites	0.85	0.02	ART3	-357	N
rs11697186	Response_to_hepatitis_C_treatment	Viral_disease	0.85	0.11	RP5-1187M17.10	-31	N
rs13394720	HIV_progression	Viral_disease	0.90	0.02	AC019221.4	-239	N
rs2885805	Cytomegalovirus_antibody_response	Viral_disease	0.87	0.00	C1orf88	471	N
rs9267665	Hepatitis_B_vaccine_response	Viral_disease	0.94	0.07	UQCRHP1	-289	N

Table S7. Genes correlated with distal DHSs harboring GWAS SNPs (Part II)

128 trait-SNP associations (123 unique SNPs) overlapping predicted long-distance interactions established by correlation of chromatin accessibility (r) in DHSs in 46 additional cell/tissue types. Cor_gene_name represents the most-correlated gene; Dist, distance to gene in kb. "Adjacent?", whether the highest-correlated gene is an adjacent gene.

SNP	Disease or trait	Trait Category	r	Cor_gene_name	Dist	Adjacent?
rs62209	Alzheimers_disease_late_onset	Aging	0.94	RP11-271F18.2	496	N
rs4938933	Alzheimers_disease_late_onset	Aging	0.89	MS4A3	201	N
rs2619566	Amyotrophic_lateral_sclerosis-age_of_onset	Aging	0.88	CNTN4	307	N
rs9664222	Longevity	Aging	0.74	RP11-57C13.5	62	N
rs1036819	Longevity	Aging	0.87	RP11-513H8.1	279	N
rs947211	Parkinsons_disease	Aging	0.96	SLC26A9	147	N
rs6599388	Parkinsons_disease	Aging	0.84	PDE6B	294	N
rs6599389	Parkinsons_disease	Aging	0.84	PDE6B	294	N
rs11248060	Parkinsons_disease	Aging	0.99	SPON2	231	N
rs4698412	Parkinsons_disease	Aging	0.70	PROM1	349	N
rs10121009	Parkinsons_disease	Aging	0.83	RP11-156G14.4	180	N
rs10767971	Parkinsons_disease_age_of_onset	Aging	0.83	PIGCP1	202	N
rs17565841	Parkinsons_disease_age_of_onset	Aging	0.74	AC090696.2	23	N
rs13010713	Celiac_disease	Autoimmune_disease	0.74	AC104820.2	7	N
rs1819658	Crohns_disease	Autoimmune_disease	0.77	UBE2D1	211	N
rs762421	Crohns_disease	Autoimmune_disease	0.75	TRAPP10	105	N
rs6596075	Crohns_disease	Autoimmune_disease	0.77	RAD50	188	N
rs212388	Crohns_disease	Autoimmune_disease	0.84	RP3-393E18.1	458	N
rs212388	Crohns_disease_celiac_disease	Autoimmune_disease	0.84	RP3-393E18.1	458	N
rs9355610	Graves_disease	Autoimmune_disease	0.73	RNASET2	12	N
rs6604026	Multiple_sclerosis	Autoimmune_disease	0.89	RP4-612C19.1	357	N
rs12025416	Multiple_sclerosis	Autoimmune_disease	0.89	RP4-655J12.5	62	N
rs7090512	Multiple_sclerosis	Autoimmune_disease	0.86	RP11-414H17.2	3	N
rs4939490	Multiple_sclerosis	Autoimmune_disease	0.93	ZP1	153	N
rs2248359	Multiple_sclerosis	Autoimmune_disease	0.75	CYP24A1	1	N
rs9321490	Multiple_sclerosis	Autoimmune_disease	0.98	AHI1	149	N
rs7779014	Multiple_sclerosis	Autoimmune_disease	0.87	POR	439	N
rs17149161	Multiple_sclerosis	Autoimmune_disease	0.80	AC005077.14	274	N
rs9303277	Primary_biliary_cirrhosis	Autoimmune_disease	0.72	IKZF3	44	N
rs842636	Psoriasis	Autoimmune_disease	0.90	AC007381.2	481	N
rs743777	Rheumatoid_arthritis	Autoimmune_disease	0.89	C1QTNF6	42	N
rs1600249	Rheumatoid_arthritis	Autoimmune_disease	0.94	CTSB	359	N
rs7329174	Systemic_lupus_erythematosus	Autoimmune_disease	0.82	ELF1	1	N
rs1317209	Ulcerative_colitis	Autoimmune_disease	0.86	TMCO4	75	N
rs3024493	Ulcerative_colitis	Autoimmune_disease	0.81	RP11-534L20.4	242	N
rs8067378	Ulcerative_colitis	Autoimmune_disease	0.86	IKZF3	31	N
rs11676348	Ulcerative_colitis	Autoimmune_disease	0.81	C2orf62	219	N
rs6017342	Ulcerative_colitis	Autoimmune_disease	0.71	WISP2	282	N
rs11978267	Acute_lymphoblastic_leukemia_childhood	Cancer	0.85	IKZF1	99	N
rs2380205	Breast_cancer	Cancer	0.92	GDI2	58	N
rs2981579	Breast_cancer	Cancer	0.96	TACC2	411	N
rs1219648	Breast_cancer	Cancer	0.79	FGFR2	7	N
rs10937405	Lung_adenocarcinoma	Cancer	0.84	TPRG1	451	N
rs7521902	Ovarian_cancer	Cancer	0.93	HSPG2	268	N
rs9311171	Prostate_cancer	Cancer	0.80	DLEC1	105	N
rs10263935	Aortic_root_size	Cardiovascular	0.75	RABGEF1	163	N
rs17375901	Atrial_fibrillation	Cardiovascular	0.99	NPPA	55	N
rs1320448	Cardiac_hypertrophy	Cardiovascular	0.98	COL17A1	0	N
rs216172	Coronary_heart_disease	Cardiovascular	0.78	MNT	178	N
rs7651039	Coronary_heart_disease	Cardiovascular	0.84	CAPN7	360	N
rs17577085	Coronary_heart_disease	Cardiovascular	0.83	ARHGAP26	423	N
rs17609940	Coronary_heart_disease	Cardiovascular	0.83	DEF6	241	N
rs6601530	Internal_carotid_intimal_medial_thickness	Cardiovascular	0.71	SOX7	83	N
rs499818	Major_CVD	Cardiovascular	0.81	GFOD1	78	N

SNP	Disease or trait	Trait Category	r	Cor_gene_name	Dist	Adjacent?
rs11748327	Myocardial_infarction	Cardiovascular	0.88	CTD-2287N17.1	16	N
rs10757278	Myocardial_infarction	Cardiovascular	0.85	CDKN2B-AS1	11	N
rs3807989	PR_interval	Cardiovascular	0.82	ST7	469	N
rs17421627	Retinal_vascular_caliber	Cardiovascular	0.89	CTC-547D20.1	127	N
rs225717	Retinal_vascular_caliber	Cardiovascular	0.90	GPR126	75	N
rs4975709	Stroke	Cardiovascular	0.77	CTD-2194D22.2	20	N
rs10829156	Sudden_cardiac_arrest	Cardiovascular	0.71	RP11-288D15.2	390	N
rs16866933	Sudden_cardiac_arrest	Cardiovascular	0.85	AC092642.1	111	N
rs7042864	Tonometry	Cardiovascular	0.88	RP11-272G11.1	143	N
rs4948088	Type_1_diabetes	Diabetes	0.71	RP4-724E13.2	93	N
rs3788013	Type_1_diabetes_autoantibodies	Diabetes	0.86	AP001625.6	140	N
rs743777	Type_1_diabetes_autoantibodies	Diabetes	0.89	C1QTNF6	42	N
rs7901695	Type_2_diabetes	Diabetes	0.71	TCF7L2	157	N
rs2383208	Type_2_diabetes	Diabetes	0.77	RP11-70L8.3	329	N
rs2472297	Coffee_consumption	Drug_metabolism	1.00	CYP11A1	395	N
rs6588480	Response_to_statin_therapy-chol_sum	Drug_metabolism	0.76	HSPB11	417	N
rs9305406	Response_to_statin_therapy-SM	Drug_metabolism	0.71	KRTAP23-1	336	N
rs17135859	F-cell_distribution	Hematological_parameters	0.79	YTHDC2	71	N
rs17342717	Mean_corpuscular_hemoglobin	Hematological_parameters	0.74	RP3-501N12.3	140	N
rs131794	Mean_corpuscular_volume	Hematological_parameters	0.80	NCAPH2	17	N
rs172629	Mean_corpuscular_volume	Hematological_parameters	0.72	RP11-601I15.1	321	N
rs12485738	Mean_platelet_volume	Hematological_parameters	0.76	C3orf63	163	N
rs8022206	Platelet_count	Hematological_parameters	0.83	RAD51L1	233	N
rs441460	Platelet_count	Hematological_parameters	0.93	RP3-522P13.3	285	N
rs11611647	Red_blood_cell_count	Hematological_parameters	0.86	DYRK4	342	N
rs7805747	Chronic_kidney_disease	Kidney_lung_liver	0.86	RP13-452N2.1	96	N
rs10786284	ADHD	Neurological_behavioral	0.88	BLNK	147	N
rs1859156	ADHD	Neurological_behavioral	0.93	PDLIM5	329	N
rs12020569	Alcohol_dependence	Neurological_behavioral	0.88	RPS3AP44	437	N
rs12282742	Bipolar_disorder_and_schizophrenia	Neurological_behavioral	0.80	RP11-113D6.8	70	N
rs17197037	Bipolar_disorder	Neurological_behavioral	0.82	METTL3	248	N
rs6990255	Bipolar_disorder	Neurological_behavioral	0.90	RP1-273G13.3	256	N
rs1574192	Brain_imaging_in_schizophrenia_interaction	Neurological_behavioral	0.95	KIF1A	429	N
rs9442235	Cognitive_performance-PC1	Neurological_behavioral	0.84	RP11-169K16.4	315	N
rs16880441	Conduct_disorder_interaction	Neurological_behavioral	0.95	ACTBP8	31	N
rs332034	Conduct_disorder_interaction	Neurological_behavioral	0.92	RP11-115J16.1	389	N
rs3827730	Depression_and_alcohol_dependence	Neurological_behavioral	0.76	RP11-183G22.3	57	N
rs12042938	DISC1	Neurological_behavioral	0.91	C1orf131	455	N
rs1869901	Schizophrenia	Neurological_behavioral	0.70	IVD	111	N
rs8005962	Tuberculosis	Parasitic_bacterial_disease	0.84	TCL6	104	N
rs6545883	Tuberculosis	Parasitic_bacterial_disease	0.71	USP34	323	N
rs9990343	Brain_structure	Quantitative_traits	0.94	CXCR6	357	N
rs17646946	Hair_curl	Quantitative_traits	0.99	TCHHL1	1	N
rs17318596	Height	Quantitative_traits	0.71	BCKDHA	20	N
rs3791679	Height	Quantitative_traits	0.81	CCDC88A	470	N
rs6724465	Height	Quantitative_traits	0.94	NHEJ1	46	N
rs12658202	Height	Quantitative_traits	0.97	FAM114A2	430	N
rs6569648	Height	Quantitative_traits	0.74	ARHGAP18	430	N
rs6570507	Height	Quantitative_traits	0.79	GPR126	58	N
rs6611365	Optic_disc_size_disc	Quantitative_traits	0.70	CTD-2522E6.4	157	N
rs9386463	Primary_tooth_development_time_to_first_tooth_eruption	Quantitative_traits	0.99	PRDM1	453	N
rs1572050	Renal_sinus_fat	Quantitative_traits	0.80	RP11-23P11.2	30	N
rs9315632	Waist-hip_ratio	Quantitative_traits	0.73	C13orf23	70	N
rs1055144	Waist-hip_ratio	Quantitative_traits	0.70	CTA-242H14.1	141	N
rs9594738	Bone_mineral_density-hip	Radiographic_parameters	0.90	FABP3P2	8	N
rs9594738	Bone_mineral_density-spine	Radiographic_parameters	0.90	FABP3P2	8	N
rs4729260	Bone_mineral_density-spine	Radiographic_parameters	0.85	SHFM1	209	N
rs10492681	Alanine_aminotransferase	Serum_metabolites	0.76	RP11-518D7.1	52	N
rs2280401	Albumin	Serum_metabolites	0.71	AKT1S1	379	N
rs16856332	Alkaline_phosphatase	Serum_metabolites	0.77	AC007556.2	129	N
rs6742078	Bilirubin	Serum_metabolites	0.94	TRPM8	168	N
rs7953249	DG7_glycan	Serum_metabolites	0.75	HNF1A	12	N
rs17342717	Ferritin	Serum_metabolites	0.74	RP3-501N12.3	140	N
rs12029080	Fibrin-D-dimer_levels	Serum_metabolites	0.79	CNN3	340	N
rs1490453	Fibrinogen	Serum_metabolites	0.75	RP11-655B23.1	127	N
rs7998202	HbA1C	Serum_metabolites	0.72	MCF2L	409	N

SNP	Disease or trait	Trait Category	r	Cor_gene_name	Dist	Adjacent?
rs7499892	HDL_cholesterol	Serum_metabolites	0.84	NLRC5	100	N
rs2083637	HDL_cholesterol	Serum_metabolites	0.77	CSGALNACT1	404	N
rs13702	HDL_cholesterol_triglycerides	Serum_metabolites	0.84	CSGALNACT1	363	N
rs9303029	IGF1-free	Serum_metabolites	0.85	FOXK2	69	N
rs7577642	IL6R	Serum_metabolites	0.81	SH2D6	43	N
rs591044	Insulin_resistance	Serum_metabolites	0.88	RP11-259P1.1	45	N
rs2280401	Protein-total	Serum_metabolites	0.71	AKT1S1	379	N
rs236918	Transferrin_receptor	Serum_metabolites	0.77	PAFAH1B2	64	N

Table S8. Search terms for association of GO Biological Processes with Disease/Trait classes

GO search terms were used to associate transcription factors with disease/trait.

Disease class	Controlled vocabulary used for GO queries
Aging	"aging", "death"
Autoimmune	"immune", "interleukin", "B cell", "T-cell", "B-cell", "T-cell", "cytokine", "NF-kappa", "leukocyte", "lymphocyte", "interferon"
Cancer	"proliferation", "tumor", "P53", "apopto", "death", "RAS"
Cardiovascular	"heart", "cardio", "angio", "artery", "cardiac", "circulat", "vascu"
Diabetes	"glucose", "insulin", "glycolysis", "glycogen", "hexose", "gluconeogenesis", "pancreatic B cell", "keto"
Drug	"xenobiotic", "drug"
Hematological	"blood", "hema", "hemo", "myeloid"
Kidney, Lung, Liver	"kidney", "neph", "lung", "hepatic", "liver", "hepatico", "bile", "urogen", "renal", "urete", "pulmon"
Lipid	"lipid", "sterol", "glyceride", "phosphatidyl", "sphingo", "acylglycerol", "icosanoid", "steroid", "adipose", "fat"
Neural	"neur", "brain", "action potential", "astrocyte", "axon", "hindbrain", "hippocampus", "spinal", "nervous", "oligodendrocyte", "cerebellar", "perception", "nerve", "glial", "glio"

Table S9. GWAS SNPs specifying allelic chromatin states.

GWAS SNPs overlapping DHSs showing significant allelic imbalance at least one cell type or tissue. Each row represents a single GWAS SNP; coordinates are hg19, 0-indexed. ref_count, alt_count, number of reads in heterozygous cell types including each allele. P-value, binomial test for significant imbalance between alleles; NA indicates lack of read depth to perform test. q-value, FDR-adjusted significance of test. GWAS_traits, comma-separated list of traits associated with SNP.

chrom	chromStart	chromEnd	dbSNP_ID	ref_allele	alt_allele	ref_count	alt_count	P-value	q-value	GWAS_traits
chr1	2069680	2069681	rs3753242	C	T	14	11	6.90E-01	9.20E-01	Olanzapine Schizophrenia neurocognition
chr1	2513215	2513216	rs734999	C	T	95	99	8.30E-01	1.00E+00	Ulcerative colitis
chr1	6279369	6279370	rs846111	G	C	6	5	NA	NA	QT interval
chr1	8021972	8021973	rs35675666	G	T	576	587	7.47E-01	9.50E-01	Ulcerative colitis
chr1	10796865	10796866	rs880315	T	C	34	28	5.26E-01	8.32E-01	Diastolic blood pressure, Systolic blood pressure
chr1	11852515	11852516	rs17375901	C	T	4	6	NA	NA	Atrial fibrillation
chr1	11856377	11856378	rs1801133	G	A	46	48	9.18E-01	1.00E+00	Plasma homocysteine
chr1	11862777	11862778	rs17367504	A	G	12	20	2.15E-01	5.15E-01	Blood pressure, Systolic blood pressure
chr1	11899032	11899033	rs1023252	G	T	26	34	3.66E-01	6.81E-01	Natriuretic peptide
chr1	16393356	16393357	rs9442235	T	G	6	8	NA	NA	Cognitive performance-PC1
chr1	17216330	17216331	rs6586513	C	A	55	48	5.55E-01	8.39E-01	Allergic rhinitis
chr1	17331675	17331676	rs3738814	A	G	29	18	1.44E-01	4.09E-01	Height
chr1	18795254	18795255	rs3007729	T	C	18	9	1.22E-01	3.66E-01	Diabetic retinopathy
chr1	20140035	20140036	rs1317209	G	A	49	36	1.93E-01	4.87E-01	Ulcerative colitis
chr1	21766452	21766453	rs1976403	A	C	176	164	5.51E-01	8.37E-01	Alkaline phosphatase
chr1	25768936	25768937	rs10903129	A	G	24	32	3.50E-01	6.69E-01	Cholesterol, Erythrocyte sedimentation rate
chr1	40064960	40064961	rs12037222	G	A	10	8	NA	NA	C-reactive protein
chr1	40132794	40132795	rs873917	T	G	33	35	9.04E-01	1.00E+00	Amyotrophic lateral sclerosis-susceptibility
chr1	50937847	50937848	rs3827730	T	C	7	7	NA	NA	Depression and alcohol dependence
chr1	53978118	53978119	rs6588480	A	G	213	157	4.18E-03	2.98E-02	Response to statin therapy-chol sum
chr1	56962820	56962821	rs17114036	A	G	14	63	1.41E-08	5.16E-07	Coronary heart disease
chr1	59162147	59162148	rs2811893	T	C	5	11	NA	NA	Diabetic retinopathy
chr1	63191776	63191777	rs12130333	C	T	11	18	2.65E-01	5.75E-01	Triglycerides
chr1	63591128	63591129	rs10493340	A	G	5	8	NA	NA	Systolic blood pressure
chr1	64108770	64108771	rs2269241	T	C	7	4	NA	NA	Type 1 diabetes
chr1	65992624	65992625	rs1751492	C	T	11	4	NA	NA	Soluble leptin receptor levels
chr1	66089781	66089782	rs6700896	C	T	86	39	3.18E-05	4.42E-04	C-reactive protein
chr1	66310864	66310865	rs10493389	T	C	182	130	3.82E-03	2.75E-02	Echocardiographic traits-LAD
chr1	70921172	70921173	rs10458561	G	A	13	4	NA	NA	Risperidone influence on QT interval
chr1	89146233	89146234	rs12145922	C	A	14	20	3.92E-01	7.19E-01	Gamma glutamyl transferase
chr1	91569688	91569689	rs164898	G	A	6	7	NA	NA	Waist-hip ratio
chr1	92077096	92077097	rs1192415	G	A	5	12	NA	NA	Optic disc parameters, Optic disc size disc
chr1	93303602	93303603	rs6604026	T	C	14	27	5.96E-02	2.22E-01	Multiple sclerosis
chr1	95053352	95053353	rs12029080	T	G	14	9	4.05E-01	7.32E-01	Fibrin-D-dimer levels
chr1	109817191	109817192	rs7528419	A	G	3	8	NA	NA	Lipoprotein-associated phospholipase A2 activity and mass
chr1	109817589	109817590	rs12740374	G	T	134	296	4.00E-15	3.79E-13	LDL cholesterol
chr1	109818529	109818530	rs646776	C	T	11	11	1.00E+00	1.00E+00	Cholesterol, Coronary heart disease, LDL cholesterol, Myocardial infarction, Progranulin, Response to statin therapy-chol sum

chrom	chromStart	chromEnd	dbSNP_ID	ref_allele	alt_allele	ref_count	alt_count	P-value	q-value	GWAS_traits
chr1	111417914	111417915	rs2885805	C	A	28	28	1.00E+00	1.00E+00	Cytomegalovirus antibody response
chr1	111684275	111684276	rs1335645	A	G	6	4	NA	NA	Gamma glutamyl transferase
chr1	113216542	113216543	rs2932538	A	G	48	42	5.98E-01	8.50E-01	Blood pressure, Diastolic blood pressure, Systolic blood pressure
chr1	117100956	117100957	rs1335532	A	G	29	36	4.57E-01	7.72E-01	Multiple sclerosis
chr1	118868404	118868405	rs17038182	G	C	57	38	6.42E-02	2.35E-01	Height
chr1	119503842	119503843	rs984222	C	G	12	15	7.01E-01	9.26E-01	Waist-hip ratio
chr1	121200489	121200490	rs6600671	T	G	10	4	NA	NA	Hip geometry
chr1	145723644	145723645	rs1967017	T	C	161	115	6.65E-03	4.31E-02	Serum urate
chr1	152083324	152083325	rs11803731	A	T	37	47	3.26E-01	6.43E-01	Hair morphology
chr1	153774275	153774276	rs4434872	T	C	6	5	NA	NA	Conduct disorder symptom count
chr1	154418878	154418879	rs4537545	C	T	699	609	1.28E-02	7.07E-02	C-reactive protein
chr1	154426263	154426264	rs4129267	C	T	556	373	1.92E-09	8.03E-08	Asthma, C-reactive protein, FEF, IL6R
chr1	155162066	155162067	rs4072037	C	T	8	15	2.10E-01	5.13E-01	Gastric cancer, Serum magnesium levels
chr1	157670815	157670816	rs7528684	A	G	12	30	7.92E-03	4.89E-02	Type 1 diabetes autoantibodies
chr1	159647987	159647988	rs12093699	G	A	5	5	NA	NA	C-reactive protein
chr1	162033889	162033890	rs12143842	C	T	3	7	NA	NA	QT interval
chr1	162112909	162112910	rs16857031	C	G	21	27	4.71E-01	7.86E-01	QT interval
chr1	164739170	164739171	rs6670655	T	C	41	50	4.02E-01	7.32E-01	Height
chr1	165403902	165403903	rs285480	G	A	7	3	NA	NA	Bipolar disorder personality
chr1	167408669	167408670	rs840016	C	T	95	81	3.27E-01	6.43E-01	ZKPQ-sensation seeking
chr1	167411383	167411384	rs864537	A	G	6	4	NA	NA	Rheumatoid arthritis
chr1	169580289	169580290	rs2235302	C	T	10	17	2.48E-01	5.54E-01	Celiac disease, Rheumatoid arthritis celiac disease
chr1	169608916	169608917	rs1569476	C	T	6	6	NA	NA	P-selectin
chr1	173301515	173301516	rs1342038	G	A	180	112	8.24E-05	1.00E-03	Alzheimers Total ventricular volume
chr1	177913518	177913519	rs10913469	T	C	14	10	5.41E-01	8.34E-01	Diabetic retinopathy
chr1	183176429	183176430	rs525410	A	G	8	11	NA	NA	BMI, Weight
chr1	183852913	183852914	rs10797919	G	C	279	205	8.88E-04	7.97E-03	Systemic lupus erythematosus
chr1	184020944	184020945	rs2274432	G	A	140	147	7.23E-01	9.37E-01	ADHD
chr1	200007431	200007432	rs3790844	A	G	12	15	7.01E-01	9.26E-01	Height
chr1	200960306	200960307	rs2297909	G	A	4	14	NA	NA	Pancreatic cancer
chr1	203155881	203155882	rs4950928	G	C	50	74	3.84E-02	1.68E-01	Ankylosing spondylitis
chr1	204112983	204112984	rs3795578	G	A	22	4	5.34E-04	5.19E-03	YKL-40 levels
chr1	205235989	205235990	rs1668873	G	A	5	5	NA	NA	Acetaminophen hepatotoxicity
chr1	205676262	205676263	rs12748961	T	C	3	7	NA	NA	Mean platelet volume
chr1	206939903	206939904	rs3024505	G	A	158	163	8.23E-01	1.00E+00	Basophil count
chr1	206943967	206943968	rs3024493	C	A	50	63	2.59E-01	5.73E-01	Crohns disease, Type 1 diabetes, Type 1 diabetes autoantibodies, Ulcerative colitis
chr1	206944644	206944645	rs1518111	T	C	33	31	9.01E-01	1.00E+00	Ulcerative colitis
chr1	207872289	207872290	rs7527798	T	C	7	27	8.21E-04	7.74E-03	Behcets disease
chr1	218860067	218860068	rs993925	C	T	96	62	8.45E-03	5.09E-02	Erythrocyte sedimentation rate
chr1	219750716	219750717	rs4846567	G	T	12	29	1.15E-02	6.40E-02	FEV1/FVC
chr1	222164947	222164948	rs6687758	A	G	19	24	5.42E-01	8.34E-01	Waist-hip ratio
chr1	228997834	228997835	rs801114	T	G	7	5	NA	NA	Colorectal cancer, Progressive supranuclear palsy
chr1	230294915	230294916	rs2144300	C	T	96	154	2.95E-04	3.08E-03	Basal cell carcinoma
chr1	234984987	234984988	rs2069084	G	A	8	9	NA	NA	HDL cholesterol, Triglycerides
chr1	236313126	236313127	rs7520258	T	C	5	7	NA	NA	HIV progression
chr1	244173871	244173872	rs10927101	C	A	7	4	NA	NA	Working memory
chr1	247601594	247601595	rs12239046	T	C	10	14	5.41E-01	8.34E-01	Diabetic retinopathy
chr10	1156164	1156165	rs10794720	T	C	150	125	1.48E-01	4.15E-01	C-reactive protein
chr10	3284006	3284007	rs2764980	A	G	34	32	9.02E-01	1.00E+00	Creatinine
chr10	4719795	4719796	rs1391511	A	G	60	51	4.48E-01	7.71E-01	ADHD
chr10	5886733	5886734	rs2380205	C	T	83	88	7.60E-01	9.58E-01	Neonatal lupus
chr10	6110828	6110829	rs7090512	C	T	8	3	NA	NA	Breast cancer
chr10	6390449	6390450	rs947474	G	A	19	7	2.90E-02	1.35E-01	Multiple sclerosis
chr10	8093033	8093034	rs501764	G	T	49	53	7.67E-01	9.65E-01	Type 1 diabetes
chr10	11000338	11000339	rs62209	G	T	24	24	1.00E+00	1.00E+00	Hodgkins lymphoma
chr10	13155725	13155726	rs1561570	T	C	5	5	NA	NA	Alzheimers disease late onset
chr10										Pagets disease

chrom	chromStart	chromEnd	dbSNP_ID	ref_allele	alt_allele	ref_count	alt_count	P-value	q-value	GWAS_traits
chr10	13755543	13755544	rs1541010	C	T	34	38	7.24E-01	9.37E-01	RR interval
chr10	17130692	17130693	rs11254363	A	G	89	167	1.24E-06	2.51E-05	Vitamin B12
chr10	18727958	18727959	rs12258967	C	G	38	37	1.00E+00	1.00E+00	Blood pressure
chr10	18950554	18950555	rs10829156	T	C	8	5	NA	NA	Sudden cardiac arrest
chr10	21830103	21830104	rs11012732	A	G	6	12	NA	NA	Meningioma
chr10	30096627	30096628	rs2986971	A	G	503	319	1.38E-10	7.34E-09	NAFLDH
chr10	30316071	30316072	rs3739998	C	G	80	204	1.18E-13	9.84E-12	Coronary heart disease
chr10	34088052	34088053	rs1412115	T	C	7	8	NA	NA	Schizophrenia
chr10	44775823	44775824	rs1746048	C	T	55	109	2.99E-05	4.37E-04	Coronary heart disease, Myocardial infarction
chr10	45123378	45123379	rs11239177	T	C	78	162	6.25E-08	2.03E-06	Bipolar disorder and schizophrenia
chr10	51549495	51549496	rs10993994	T	C	7	10	NA	NA	PSA, Prostate cancer
chr10	54223976	54223977	rs1733724	A	G	6	5	NA	NA	QRS duration
chr10	59189177	59189178	rs4462262	T	C	16	4	NA	NA	Diabetic retinopathy
chr10	63723576	63723577	rs10821936	C	T	15	15	1.00E+00	1.00E+00	Acute lymphoblastic leukemia childhood
chr10	63752158	63752159	rs7089424	T	G	151	146	8.17E-01	1.00E+00	Acute lymphoblastic leukemia childhood
chr10	64251976	64251977	rs10822013	C	T	311	226	2.44E-04	2.60E-03	Breast cancer
chr10	65133821	65133822	rs7923609	A	G	5	6	NA	NA	Alkaline phosphatase
chr10	65274926	65274927	rs10761779	A	G	108	197	3.91E-07	9.12E-06	Alkaline phosphatase
chr10	71093391	71093392	rs16926246	C	T	17	20	7.43E-01	9.49E-01	HbA1C, Hematocrit, Hemoglobin
chr10	72483009	72483010	rs1816002	T	C	50	83	5.33E-03	3.62E-02	Weight
chr10	72982984	72982985	rs16928529	G	T	89	81	5.91E-01	8.50E-01	ADHD
chr10	73849638	73849639	rs1245541	G	A	232	205	2.14E-01	5.15E-01	IGF1
chr10	80841147	80841148	rs704010	T	C	76	105	3.71E-02	1.63E-01	Breast cancer
chr10	80955066	80955067	rs703965	C	T	98	133	2.51E-02	1.20E-01	Bipolar disorder
chr10	81121695	81121696	rs2145998	T	A	120	78	3.47E-03	2.60E-02	Height
chr10	89338632	89338633	rs9664222	A	C	57	91	6.48E-03	4.25E-02	Longevity
chr10	93348119	93348120	rs1329650	G	T	93	80	3.62E-01	6.79E-01	Smoking behavior
chr10	94347829	94347830	rs6583826	G	A	6	4	NA	NA	Type 2 diabetes
chr10	94462881	94462882	rs1111875	C	T	6	6	NA	NA	Type 2 diabetes
chr10	94485210	94485211	rs2497306	A	C	6	7	NA	NA	Serum dehydroepiandrosterone
chr10	96070374	96070375	rs3781264	A	G	10	13	6.78E-01	9.10E-01	Gastric cancer
chr10	98135504	98135505	rs10786284	A	T	15	11	5.57E-01	8.39E-01	ADHD
chr10	101284236	101284237	rs4409764	T	G	28	24	6.78E-01	9.10E-01	Crohns disease
chr10	101290300	101290301	rs6584283	T	C	23	32	2.81E-01	5.92E-01	Ulcerative colitis
chr10	102501570	102501571	rs4509693	C	T	11	6	NA	NA	Alzheimers disease
chr10	104719095	104719096	rs12413409	G	A	5	6	NA	NA	Coronary heart disease, Intracranial aneurysm
chr10	104846177	104846178	rs11191548	T	C	8	4	NA	NA	Blood pressure, Diastolic blood pressure, Systolic blood pressure
chr10	104906210	104906211	rs11191580	T	C	40	66	1.48E-02	7.99E-02	Schizophrenia
chr10	105677896	105677897	rs4387287	A	C	713	707	8.73E-01	1.00E+00	Telomere length
chr10	105846073	105846074	rs1320448	A	G	5	8	NA	NA	Cardiac hypertrophy
chr10	107516351	107516352	rs17119461	T	C	12	11	1.00E+00	1.00E+00	Melanoma
chr10	116139028	116139029	rs4751674	T	C	9	8	NA	NA	Cognitive_performance-PC1
chr10	118575605	118575606	rs740363	G	A	31	30	1.00E+00	1.00E+00	Heart failure
chr10	123337334	123337335	rs2981579	A	G	22	19	7.55E-01	9.55E-01	Breast cancer
chr11	1874071	1874072	rs9076111	G	A	20	10	9.87E-02	3.20E-01	Ulcerative colitis
chr11	1941945	1941946	rs9091116	T	C	9	15	3.07E-01	6.21E-01	Breast cancer
chr11	2170142	2170143	rs1004446	G	A	6	32	2.43E-05	3.65E-04	Type 1 diabetes, Type 1 diabetes autoantibodies
chr11	2213165	2213166	rs7111341	C	T	17	11	3.45E-01	6.63E-01	Type 1 diabetes
chr11	2484802	2484803	rs2074238	T	C	13	15	8.51E-01	1.00E+00	QT interval
chr11	2858545	2858546	rs2237897	C	T	64	64	1.00E+00	1.00E+00	Type 2 diabetes
chr11	2953722	2953723	rs3847646	C	T	6	4	NA	NA	Serum calcium
chr11	10350537	10350538	rs7129220	G	A	142	146	8.60E-01	1.00E+00	Blood pressure, Diastolic blood pressure, Systolic blood pressure
chr11	10669227	10669228	rs7940646	T	C	185	177	7.13E-01	9.36E-01	Platelet aggregation-ADP
chr11	16902267	16902268	rs381815	C	T	4	7	NA	NA	Blood pressure, Systolic blood pressure
chr11	16917218	16917219	rs11024074	T	C	20	28	3.12E-01	6.25E-01	Diastolic blood pressure
chr11	17409571	17409572	rs5219	T	C	6	6	NA	NA	Type 2 diabetes
chr11	18285773	18285774	rs4638289	A	T	17	27	1.74E-01	4.58E-01	Atherosclerosis

chrom	chromStart	chromEnd	dbSNP_ID	ref_allele	alt_allele	ref_count	alt_count	P-value	q-value	GWAS_traits
chr11	22829756	22829757	rs7111546	C	T	37	25	1.62E-01	4.40E-01	Dialysis-related mortality
chr11	46722220	46722221	rs7932354	T	C	2763	2984	3.55E-03	2.63E-02	Bone mineral density-hip
chr11	47286289	47286290	rs7120118	T	C	60	123	3.71E-06	6.38E-05	HDL cholesterol
chr11	47293798	47293799	rs10501320	G	C	21	14	3.11E-01	6.23E-01	Proinsulin levels
chr11	47663048	47663049	rs10838738	A	G	8	17	1.08E-01	3.38E-01	BMI
chr11	60760611	60760612	rs17824933	C	G	31	29	8.97E-01	1.00E+00	Multiple sclerosis
chr11	61551355	61551356	rs174535	T	C	5	15	NA	NA	Serum polyunsaturated fatty acids
chr11	61560080	61560081	rs174538	G	A	1153	1284	7.96E-03	4.89E-02	Serum polyunsaturated fatty acids
chr11	61597971	61597972	rs1535	A	G	30	48	5.35E-02	2.06E-01	HDL cholesterol, Response to statin therapy-braces, Serum polyunsaturated fatty acids
chr11	61609749	61609750	rs174583	C	T	15	20	5.00E-01	8.11E-01	Response to statin therapy-Triglyceride sum
chr11	61655304	61655305	rs1000778	A	G	3	7	NA	NA	Sphingolipid concentrations
chr11	61815802	61815803	rs4963452	T	C	20	67	4.31E-07	9.67E-06	Serum polyunsaturated fatty acids
chr11	63963946	63963947	rs11607165	T	G	7	11	NA	NA	ADHD
chr11	64334113	64334114	rs2078267	C	T	339	335	8.78E-01	1.00E+00	Gout, Serum urate
chr11	64546390	64546391	rs606458	C	T	1594	1658	2.62E-01	5.75E-01	Serum urate
chr11	65249144	65249145	rs17146964	A	G	109	54	1.98E-05	3.04E-04	Optic vertical cup-disc ratio
chr11	69234493	69234494	rs4980785	C	T	56	39	1.00E-01	3.20E-01	Renal cell carcinoma
chr11	72432984	72432985	rs11603334	G	A	69	55	2.43E-01	5.54E-01	Proinsulin levels
chr11	72433097	72433098	rs1552224	A	C	48	57	4.35E-01	7.65E-01	Type 2 diabetes
chr11	75276177	75276178	rs606452	A	C	47	39	4.51E-01	7.72E-01	Height
chr11	75282051	75282052	rs634552	T	G	44	30	1.30E-01	3.86E-01	Height
chr11	75909618	75909619	rs4944092	A	G	12	4	NA	NA	PR interval
chr11	85800278	85800279	rs561655	G	A	16	22	4.18E-01	7.46E-01	Alzheimers disease late onset
chr11	88911695	88911696	rs1042602	C	A	9	4	NA	NA	Freckles, Skin pigmentation
chr11	92673827	92673828	rs1387153	C	T	7	3	NA	NA	Blood pressure fasting glucose, Fasting plasma glucose, HDL cholesterol fasting glucose, HbA1C, Type 2 diabetes
chr11	95311421	95311422	rs4409785	T	C	11	43	1.40E-05	2.27E-04	Multiple sclerosis
chr11	106330161	106330162	rs10895959	G	A	13	9	5.23E-01	8.32E-01	ADHD-Inattentive symptoms
chr11	109995943	109995944	rs746463	C	T	5	6	NA	NA	Exercise treadmill test traits
chr11	114191015	114191016	rs2847476	A	G	3	7	NA	NA	Volumetric brain MRI
chr11	116603723	116603724	rs12272004	C	A	5	5	NA	NA	Alpha-tocopherol, Cholesterol, LDL cholesterol, Triglycerides
chr11	116613659	116613660	rs12280753	C	T	157	148	6.47E-01	8.94E-01	Triglycerides
chr11	116648916	116648917	rs964184	G	C	16	18	8.64E-01	1.00E+00	Alpha-tocopherol, Coronary heart disease, HDL cholesterol, Hypertriglyceridemia, Lipoprotein-associated phospholipase A2 activity and mass, Triglycerides
chr11	116732511	116732512	rs2075292	G	T	261	315	2.44E-02	1.20E-01	Triglycerides
chr11	118145685	118145686	rs17122021	C	T	8	4	NA	NA	Pain
chr11	118743771	118743772	rs6421571	T	C	4	12	NA	NA	Primary biliary cirrhosis
chr11	119099905	119099906	rs4938642	G	C	13	4	NA	NA	Platelet count
chr11	122030189	122030190	rs577948	A	G	264	295	1.90E-01	4.82E-01	Myopia pathological
chr11	125178402	125178403	rs10893366	C	T	9	5	NA	NA	Alcohol dependence
chr11	125461708	125461709	rs548181	A	G	28	39	2.22E-01	5.25E-01	Schizophrenia
chr11	128206409	128206410	rs6590322	C	T	154	129	1.54E-01	4.25E-01	Hippocampal atrophy
chr11	128380973	128380974	rs11221332	C	T	4	6	NA	NA	Celiac disease
chr11	130273229	130273230	rs11222084	A	T	558	514	1.79E-01	4.67E-01	Blood pressure
chr11	131320069	131320070	rs1550976	T	C	15	4	NA	NA	Asperger disorder
chr12	783483	783484	rs12425791	G	A	27	15	8.84E-02	2.97E-01	Stroke
chr12	3814237	3814238	rs10848911	G	A	74	49	3.00E-02	1.39E-01	Male infertility
chr12	6289218	6289219	rs1558324	G	A	31	26	5.97E-01	8.50E-01	Mean platelet volume
chr12	6291092	6291093	rs7342306	G	A	71	89	1.79E-01	4.67E-01	Platelet count
chr12	6580581	6580582	rs10492096	A	G	29	35	5.32E-01	8.32E-01	Hip geometry
chr12	9876090	9876091	rs10466829	G	A	17	10	2.48E-01	5.54E-01	Multiple sclerosis
chr12	11855623	11855624	rs2187642	A	C	5	7	NA	NA	Height
chr12	15930804	15930805	rs17415853	T	C	5	13	NA	NA	Ferritin

chrom	chromStart	chromEnd	dbSNP_ID	ref_allele	alt_allele	ref_count	alt_count	P-value	q-value	GWAS_traits
chr12	20521653	20521654	rs10841496	C	A	280	242	9.63E-02	3.18E-01	Male infertility
chr12	24389659	24389660	rs1464500	G	T	11	17	3.45E-01	6.63E-01	Response to antipsychotic therapy perphenazine-HDL
chr12	29435479	29435480	rs2015599	G	A	28	23	5.76E-01	8.50E-01	Mean platelet volume
chr12	47639525	47639526	rs11610206	T	C	3	11	NA	NA	Alzheimers disease
chr12	49218170	49218171	rs2070615	A	G	46	37	3.80E-01	7.02E-01	Bipolar disorder
chr12	51155662	51155663	rs11169552	C	T	9	4	NA	NA	Colorectal cancer
chr12	53273903	53273904	rs902774	G	A	343	261	8.48E-04	7.86E-03	Prostate cancer
chr12	53731890	53731891	rs10876432	G	A	10	9	NA	NA	Bone mineral density-spine
chr12	54342683	54342684	rs1443512	A	C	9	11	NA	NA	Waist-hip ratio
chr12	57503774	57503775	rs167769	C	T	13	7	NA	NA	Eosinophilic esophagitis pediatric
chr12	57527282	57527283	rs1172113	T	C	166	104	1.94E-04	2.11E-03	FEV1/FVC, Migrane
chr12	58162738	58162739	rs703842	A	G	10	3	NA	NA	Multiple sclerosis
chr12	66227256	66227257	rs7979673	C	T	5	5	NA	NA	Height
chr12	66358346	66358347	rs1042725	C	T	7	5	NA	NA	Height
chr12	68500074	68500075	rs7134599	G	A	83	90	6.48E-01	8.94E-01	Ulcerative colitis
chr12	69827657	69827658	rs10748128	G	T	39	20	1.83E-02	9.48E-02	Height
chr12	71577100	71577101	rs1495377	G	C	16	14	8.56E-01	1.00E+00	Type 2 diabetes
chr12	88890670	88890671	rs995030	A	G	39	42	8.24E-01	1.00E+00	Testicular cancer
chr12	88953958	88953959	rs4474514	G	A	6	18	2.27E-02	1.13E-01	Testicular cancer
chr12	89328334	89328335	rs12821256	T	C	73	94	1.21E-01	3.66E-01	Hair color-blond vs. brown
chr12	90013088	90013089	rs2681492	T	C	71	76	7.42E-01	9.49E-01	Systolic blood pressure
chr12	93550300	93550301	rs1836127	G	T	9	6	NA	NA	NAFLDH
chr12	99961348	99961349	rs2373011	C	G	4	11	NA	NA	BMI, Waist circumference
chr12	108699031	108699032	rs1878022	C	T	14	18	5.97E-01	8.50E-01	Response to cisplatin
chr12	111884607	111884608	rs3184504	T	C	31	38	4.70E-01	7.86E-01	Coronary heart disease, Diastolic blood pressure, Plasma eosinophil count, Platelet count, Rheumatoid arthritis, Systolic blood pressure, Type 1 diabetes, Type 1 diabetes autoantibodies
chr12	115094259	115094260	rs11067228	A	G	139	189	6.73E-03	4.32E-02	PSA
chr12	121402931	121402932	rs7305618	C	T	14	8	2.86E-01	5.99E-01	C-reactive protein, Type 2 diabetes
chr12	121423955	121423956	rs2393791	C	T	8	7	NA	NA	Gamma glutamyl transferase
chr12	121442669	121442670	rs1169313	T	C	16	10	3.27E-01	6.43E-01	Gamma glutamyl transferase
chr12	122365582	122365583	rs7961894	C	T	13	10	6.78E-01	9.10E-01	Mean platelet volume, Platelet count
chr12	123656724	123656725	rs1790100	G	T	18	15	7.28E-01	9.39E-01	Multiple sclerosis
chr12	131576190	131576191	rs1569019	C	A	7	5	NA	NA	Height
chr13	23755126	23755127	rs4770403	G	A	17	26	2.22E-01	5.25E-01	Alcohol dependence
chr13	24205194	24205195	rs9510787	A	G	8	17	1.08E-01	3.38E-01	Nasopharyngeal carcinoma
chr13	24658355	24658356	rs9805786	G	T	92	96	8.27E-01	1.00E+00	Depression and alcohol dependence
chr13	27531266	27531267	rs17085007	T	C	24	11	4.10E-02	1.73E-01	Ulcerative colitis
chr13	27586872	27586873	rs12020569	G	C	9	13	5.23E-01	8.32E-01	Alcohol dependence
chr13	28077143	28077144	rs9512730	G	C	28	24	6.78E-01	9.10E-01	Schizophrenia
chr13	28929710	28929711	rs17086609	A	G	68	26	1.73E-05	2.73E-04	Cognitive performance-IED
chr13	33693836	33693837	rs9315204	C	T	19	11	2.00E-01	4.98E-01	Intracranial aneurysm
chr13	40350911	40350912	rs7993214	T	C	216	257	6.58E-02	2.35E-01	Psoriasis
chr13	41013976	41013977	rs9418232	T	C	24	12	6.52E-02	2.35E-01	Ulcerative colitis
chr13	41110883	41110884	rs2721051	C	T	26	30	6.89E-01	9.20E-01	Central corneal thickness
chr13	41558109	41558110	rs7329174	A	G	11	9	NA	NA	Systemic lupus erythematosus
chr13	42951448	42951449	rs9533090	C	T	8	5	NA	NA	Bone mineral density-spine
chr13	42952144	42952145	rs9594738	C	T	129	107	1.71E-01	4.53E-01	Bone mineral density-hip, Bone mineral density-spine
chr13	43032592	43032593	rs9594759	C	T	11	7	NA	NA	Bone mineral density-spine
chr13	43727848	43727849	rs1324015	A	G	13	25	7.30E-02	2.54E-01	Cognitive performance-PC1
chr13	44457924	44457925	rs3764147	A	G	22	82	2.57E-09	1.00E-07	Crohns disease, Leprosy
chr13	50841322	50841323	rs806321	C	T	7	3	NA	NA	Multiple sclerosis
chr13	51111354	51111355	rs3116602	T	G	10	12	8.32E-01	1.00E+00	Height
chr13	72347695	72347696	rs626277	A	C	17	14	7.20E-01	9.37E-01	Creatinine
chr13	74520185	74520186	rs1886512	T	A	123	160	3.22E-02	1.46E-01	QRS duration
chr13	80668873	80668874	rs9574565	T	C	16	16	1.00E+00	1.00E+00	Nonsyndromic cleft lip with or without cleft palate
chr13	93213475	93213476	rs1413191	C	T	22	21	1.00E+00	1.00E+00	HIV progression

chrom	chromStart	chromEnd	dbSNP_ID	ref_allele	alt_allele	ref_count	alt_count	P-value	q-value	GWAS_traits
chr13	98016415	98016416	rs9556711	G	A	23	13	1.32E-01	3.91E-01	Alcohol dependence, Alcohol use disorder
chr13	103867103	103867104	rs912969	C	T	4	6	NA	NA	Smoking behavior
chr13	107780315	107780316	rs1927745	G	A	5	10	NA	NA	Major depressive disorder
chr13	111030577	111030578	rs7319311	A	G	20	15	5.00E-01	8.11E-01	Bipolar disorder and schizophrenia
chr13	113760033	113760034	rs561241	T	C	9	8	NA	NA	Factor VII
chr14	21725653	21725654	rs17197037	G	A	8	3	NA	NA	Bipolar disorder
chr14	23589056	23589057	rs2239633	G	A	6	4	NA	NA	Acute lymphoblastic leukemia childhood
chr14	29249348	29249349	rs2038256	T	G	12	19	2.81E-01	5.92E-01	Multiple sclerosis-Brain Glutamate Concentrations
chr14	33282469	33282470	rs2383378	C	A	8	6	NA	NA	Anorexia nervosa
chr14	35832665	35832666	rs8016947	T	G	27	38	2.15E-01	5.15E-01	Psoriasis
chr14	50877982	50877983	rs1265879	A	G	38	56	7.90E-02	2.71E-01	Trails A
chr14	52518810	52518811	rs730532	G	A	304	239	5.28E-03	3.62E-02	FEF
chr14	54410918	54410919	rs4444235	T	C	6	7	NA	NA	Colorectal cancer
chr14	64235555	64235556	rs7157785	G	T	17	50	6.74E-05	8.37E-04	Sphingolipid concentrations
chr14	65758641	65758642	rs7159888	G	A	8	8	NA	NA	DG1 glycan
chr14	68753592	68753593	rs911263	C	T	24	47	8.55E-03	5.10E-02	Primary biliary cirrhosis
chr14	69254190	69254191	rs4902647	C	T	327	307	4.27E-01	7.58E-01	Multiple sclerosis
chr14	69278203	69278204	rs4899260	C	T	32	34	9.02E-01	1.00E+00	Celiac disease
chr14	69408696	69408697	rs2268983	G	A	768	739	4.55E-01	7.72E-01	Smoking behavior
chr14	71352647	71352648	rs36563	T	G	4	6	NA	NA	Alcohol dependence
chr14	73756298	73756299	rs10483853	A	G	3	9	NA	NA	Coronary artery calcification
chr14	75626041	75626042	rs910316	A	C	19	30	1.52E-01	4.24E-01	Height
chr14	90679909	90679910	rs8017423	T	C	8	4	NA	NA	Heart failure mortality-AA
chr14	91022184	91022185	rs17793829	C	T	6	5	NA	NA	Cytomegalovirus antibody response
chr14	95720677	95720678	rs8014194	T	A	7	8	NA	NA	Response to statin therapy-chol diff
chr14	96027152	96027153	rs8005962	C	T	7	15	1.34E-01	3.91E-01	Tuberculosis
chr14	100625901	100625902	rs7157599	C	T	31	29	8.97E-01	1.00E+00	Sudden cardiac arrest
chr14	101159415	101159416	rs7149242	T	G	133	66	2.34E-06	4.42E-05	Platelet count
chr14	101306044	101306045	rs941576	A	G	40	20	1.35E-02	7.36E-02	Type 1 diabetes
chr14	103040086	103040087	rs11628318	T	A	27	30	7.91E-01	9.87E-01	Platelet count
chr14	103572814	103572815	rs944002	A	G	7	3	NA	NA	Gamma glutamyl transferase, Mean platelet volume
chr14	105729791	105729792	rs3000073	G	A	9	5	NA	NA	Mean platelet volume
chr15	37349801	37349802	rs1568679	T	C	11	8	NA	NA	Response to antipsychotic therapy risperidone-hip circumference
chr15	38907040	38907041	rs7171171	A	G	4	6	NA	NA	Type 1 diabetes autoantibodies
chr15	39315357	39315358	rs12907914	G	C	51	66	1.95E-01	4.90E-01	Cardiac hypertrophy
chr15	43559230	43559231	rs748404	T	C	39	29	2.75E-01	5.92E-01	Lung cancer
chr15	48914774	48914775	rs1036476	T	C	22	10	5.01E-02	1.98E-01	Thoracic aortic aneurysm
chr15	49192790	49192791	rs8023445	T	C	8	3	NA	NA	Major depressive disorder
chr15	51516054	51516055	rs2899472	C	A	345	389	1.04E-01	3.31E-01	Alzheimers-AB1-42
chr15	51569409	51569410	rs2305707	A	G	8	22	1.61E-02	8.48E-02	Height
chr15	58746829	58746830	rs3825776	T	C	125	116	6.06E-01	8.57E-01	Amyotrophic lateral sclerosis
chr15	60878029	60878030	rs340005	G	A	20	18	8.71E-01	1.00E+00	Gamma glutamyl transferase
chr15	60883280	60883281	rs339969	C	A	169	100	3.08E-05	4.39E-04	Gamma glutamyl transferase
chr15	62404381	62404382	rs1436955	C	T	3	9	NA	NA	Type 2 diabetes
chr15	62433961	62433962	rs11071657	A	G	44	59	1.67E-01	4.49E-01	Fasting plasma glucose
chr15	63333723	63333724	rs3809566	A	G	61	57	7.83E-01	9.81E-01	Platelet count
chr15	63341995	63341996	rs11071720	T	C	15	7	1.34E-01	3.91E-01	Mean platelet volume
chr15	67442595	67442596	rs17293632	C	T	214	55	0.00E+00	0.00E+00	Crohns disease
chr15	67446784	67446785	rs744910	G	A	28	43	9.59E-02	3.18E-01	Asthma
chr15	67458638	67458639	rs17228212	T	C	7	8	NA	NA	Coronary heart disease
chr15	73978336	73978337	rs8038465	C	T	83	94	4.52E-01	7.72E-01	Gamma glutamyl transferase
chr15	74219581	74219582	rs3825942	G	A	331	313	4.78E-01	7.96E-01	Glaucoma-exfoliation
chr15	74229064	74229065	rs893817	G	A	12	17	4.58E-01	7.72E-01	Aortic root size
chr15	75125644	75125645	rs6495122	A	C	6	5	NA	NA	Caffeine intake, Cofee consumption, Diastolic blood pressure
chr15	75382541	75382542	rs12148488	G	T	24	22	8.83E-01	1.00E+00	Caffeine intake

chrom	chromStart	chromEnd	dbSNP_ID	ref_allele	alt_allele	ref_count	alt_count	P-value	q-value	GWAS_traits
chr15	75718668	75718669	rs8028182	G	T	11	16	4.42E-01	7.66E-01	Sudden cardiac arrest
chr15	79235445	79235446	rs3825932	T	C	6	7	NA	NA	Type 1 diabetes
chr15	80694921	80694922	rs2278702	A	T	6	14	NA	NA	Bipolar disorder
chr15	84573040	84573041	rs7183263	T	G	135	157	2.19E-01	5.22E-01	Height
chr15	89902031	89902032	rs4932217	A	C	745	834	2.51E-02	1.20E-01	Height
chr15	93179174	93179175	rs285757	T	C	428	316	4.02E-05	5.34E-04	HIV progression
chr15	94036687	94036688	rs7175404	A	G	10	15	4.24E-01	7.56E-01	ADHD
chr15	99194895	99194896	rs2871865	C	G	69	67	9.32E-01	1.00E+00	Height
chr16	11158884	11158885	rs887864	G	A	17	40	3.20E-03	2.43E-02	Allergic rhinitis
chr16	11166687	11166688	rs11865121	C	A	32	27	6.03E-01	8.55E-01	Multiple sclerosis
chr16	11179872	11179873	rs12708716	A	G	19	14	4.87E-01	8.06E-01	Type 1 diabetes, Type 1 diabetes autoantibodies
chr16	11249328	11249329	rs6498169	G	A	56	73	1.59E-01	4.35E-01	Multiple sclerosis
chr16	11641179	11641180	rs7203193	A	G	118	113	7.92E-01	9.87E-01	LDL cholesterol
chr16	11691752	11691753	rs8049607	T	C	145	158	4.91E-01	8.07E-01	QT interval
chr16	20674491	20674492	rs151222	G	C	8	4	NA	NA	Schizophrenia
chr16	28539847	28539848	rs4788084	C	T	33	33	1.00E+00	1.00E+00	Type 1 diabetes, Type 1 diabetes autoantibodies
chr16	30635658	30635659	rs7186852	A	G	9	8	NA	NA	Systemic lupus erythematosus
chr16	31313252	31313253	rs9888739	C	T	8	5	NA	NA	Systemic lupus erythematosus
chr16	50719102	50719103	rs9302752	T	C	7	4	NA	NA	Leprosy
chr16	50739581	50739582	rs17221417	C	G	21	70	2.51E-07	6.11E-06	Crohns disease
chr16	52599187	52599188	rs4784227	C	T	25	31	5.04E-01	8.11E-01	Breast cancer
chr16	53809246	53809247	rs1121980	G	A	8	12	NA	NA	BMI, Obesity early onset extreme
chr16	53813366	53813367	rs17817449	T	G	14	15	1.00E+00	1.00E+00	Obesity
chr16	53816274	53816275	rs8050136	C	A	6	12	NA	NA	Adiposity, BMI, Type 2 diabetes, Weight
chr16	54494423	54494424	rs9921518	A	G	54	91	2.67E-03	2.05E-02	Dialysis-related mortality
chr16	56969147	56969148	rs2217332	G	A	18	16	8.64E-01	1.00E+00	HDL cholesterol
chr16	57005478	57005479	rs1532624	C	A	31	34	8.04E-01	9.97E-01	Cholesterol, HDL cholesterol
chr16	57006589	57006590	rs7499892	C	T	5	5	NA	NA	HDL cholesterol
chr16	67708896	67708897	rs12449157	A	G	5	5	NA	NA	HDL cholesterol
chr16	68820945	68820946	rs9929218	G	A	250	205	3.90E-02	1.69E-01	Colorectal cancer
chr16	69135048	69135049	rs8047014	C	A	55	42	2.23E-01	5.25E-01	ADHD
chr16	69588571	69588572	rs1364063	T	C	18	8	7.55E-02	2.61E-01	Menarche
chr16	80497600	80497601	rs4581712	C	A	20	33	9.84E-02	3.20E-01	Gamma glutamyl transferase
chr16	82662267	82662268	rs4783244	G	T	6	8	NA	NA	Adiponectin
chr16	82663287	82663288	rs12051272	G	T	34	54	4.22E-02	1.77E-01	Adiponectin
chr16	87111629	87111630	rs12933233	A	G	7	3	NA	NA	Amyotrophic lateral sclerosis-age of onset
chr17	1634103	1634104	rs12150338	C	T	28	23	5.76E-01	8.50E-01	Serum calcium
chr17	2126503	2126504	rs216172	G	C	443	390	6.63E-02	2.35E-01	Coronary heart disease
chr17	2143459	2143460	rs10852932	G	T	225	266	7.09E-02	2.50E-01	Aortic root size
chr17	2216257	2216258	rs391300	T	C	21	38	3.63E-02	1.61E-01	Type 2 diabetes
chr17	2262702	2262703	rs4790333	C	T	490	432	5.61E-02	2.11E-01	Proinsulin levels
chr17	9336369	9336370	rs10521157	G	A	185	157	1.44E-01	4.09E-01	Longevity
chr17	16849138	16849139	rs4273077	A	G	14	36	2.60E-03	2.03E-02	Protein-total
chr17	27714586	27714587	rs8076739	C	T	35	19	4.02E-02	1.73E-01	Mean platelet volume
chr17	35850552	35850553	rs712039	C	T	248	213	1.13E-01	3.52E-01	Tuberculosis
chr17	36098039	36098040	rs4430796	G	A	20	47	1.31E-03	1.12E-02	Endometrial cancer, PSA, Prostate cancer, Type 2 diabetes
chr17	36101155	36101156	rs7501939	T	C	9	11	NA	NA	Prostate cancer
chr17	38051347	38051348	rs8067378	A	G	3	7	NA	NA	Ulcerative colitis
chr17	38110688	38110689	rs17609240	T	G	12	11	1.00E+00	1.00E+00	WBC count
chr17	38128647	38128648	rs3859192	C	T	61	16	2.42E-07	6.11E-06	WBC count
chr17	38156711	38156712	rs4794822	C	T	7	4	NA	NA	Neutrophil count
chr17	40514200	40514201	rs744166	A	G	16	19	7.36E-01	9.47E-01	Crohns disease, Multiple sclerosis
chr17	42193184	42193185	rs228769	G	C	250	246	8.93E-01	1.00E+00	Bone mineral density-hip, Bone mineral density-spine
chr17	45013270	45013271	rs17608766	T	C	69	61	5.39E-01	8.34E-01	Blood pressure, QRS duration, Systolic blood pressure
chr17	46357119	46357120	rs2084881	G	A	5	5	NA	NA	Ovarian cancer
chr17	46411499	46411500	rs9303542	A	G	4	6	NA	NA	Ovarian cancer
chr17	46617018	46617019	rs6504340	A	G	11	16	4.42E-01	7.66E-01	Primary tooth development number of teeth
chr17	46720564	46720565	rs2326017	C	T	5	6	NA	NA	Cognitive performance-SWM

chrom	chromStart	chromEnd	dbSNP_ID	ref_allele	alt_allele	ref_count	alt_count	P-value	q-value	GWAS_traits
chr17	46988596	46988597	rs46522	C	T	6	4	NA	NA	Strategy
chr17	47440465	47440466	rs16948048	A	G	19	24	5.42E-01	8.34E-01	Coronary heart disease
chr17	47564011	47564012	rs1035050	T	C	9	14	4.05E-01	7.32E-01	Diastolic blood pressure
chr17	56584204	56584205	rs2302189	A	C	20	13	2.96E-01	6.11E-01	Bipolar disorder
chr17	59286643	59286644	rs8068952	G	C	50	43	5.34E-01	8.32E-01	Dental caries
chr17	59456588	59456589	rs9895661	C	T	34	26	3.66E-01	6.81E-01	Optic vertical cup-disc ratio
chr17	68494991	68494992	rs17779747	G	T	26	31	5.97E-01	8.50E-01	Creatinine
chr17	73872947	73872948	rs1055129	A	G	93	89	8.24E-01	1.00E+00	QT interval
chr17	78348493	78348494	rs6565681	A	G	7	8	NA	NA	White matter hyperintensity
chr17	80408814	80408815	rs9303029	C	T	727	855	1.29E-03	1.12E-02	Moyamoya disease
chr18	2948028	2948029	rs10460009	C	T	5	6	NA	NA	IGF1-free
chr18	12821592	12821593	rs2847281	A	G	14	15	1.00E+00	1.00E+00	Type 2 diabetes
chr18	20720972	20720973	rs11082304	G	T	27	26	1.00E+00	1.00E+00	C-reactive protein
chr18	21140431	21140432	rs1805081	T	C	4	6	NA	NA	Platelet count, Smoking behavior
chr18	42439885	42439886	rs991014	C	T	26	33	4.35E-01	7.65E-01	Obesity
chr18	46453462	46453463	rs4939827	T	C	15	20	5.00E-01	8.11E-01	QRS duration
chr18	57963116	57963117	rs17773430	T	C	83	95	4.10E-01	7.39E-01	Colorectal cancer
chr19	1207237	1207238	rs3764640	G	T	146	93	7.36E-04	7.04E-03	Blood pressure
chr19	1811602	1811603	rs7250872	C	T	174	181	7.50E-01	9.50E-01	Alzheimers disease
chr19	2160528	2160529	rs3803915	C	A	32	28	6.99E-01	9.26E-01	Bipolar disorder
chr19	2275078	2275079	rs2523178	A	G	27	28	1.00E+00	1.00E+00	BMI
chr19	3428833	3428834	rs7507204	G	C	282	283	1.00E+00	1.00E+00	Height
chr19	6668971	6668972	rs1077667	C	T	6	8	NA	NA	Height
chr19	7196564	7196565	rs2115386	C	T	15	19	6.08E-01	8.57E-01	Multiple sclerosis
chr19	8644030	8644031	rs4072910	G	C	24	38	9.80E-02	3.20E-01	Diabetic retinopathy
chr19	10395682	10395683	rs5498	A	G	4	13	NA	NA	ICAM1
chr19	10397402	10397403	rs3093030	C	T	226	240	5.47E-01	8.36E-01	ICAM1
chr19	11195029	11195030	rs11668477	A	G	37	29	3.89E-01	7.17E-01	LDL cholesterol
chr19	11202305	11202306	rs6511720	G	T	130	120	5.69E-01	8.50E-01	Cardiovascular disease risk factors, Internal carotid intimal medial thickness, LDL cholesterol, Lipoprotein-associated phospholipase A2 activity and mass
chr19	11210911	11210912	rs2228671	C	T	54	51	8.45E-01	1.00E+00	Cholesterol, LDL cholesterol
chr19	12146369	12146370	rs3745672	T	C	68	75	6.16E-01	8.65E-01	Multiple sclerosis
chr19	13001546	13001547	rs11085824	A	G	425	399	3.65E-01	6.81E-01	Mean corpuscular hemoglobin
chr19	17590280	17590281	rs11666579	G	T	289	330	9.94E-02	3.20E-01	Response to antidepressants genotype x drug
chr19	18304699	18304700	rs874628	A	G	44	47	8.34E-01	1.00E+00	Pain
chr19	18817902	18817903	rs10423674	C	A	40	8	3.31E-06	5.85E-05	Bipolar disorder
chr19	21666209	21666210	rs2562456	C	T	7	18	4.33E-02	1.78E-01	Lysine-valine ratio
chr19	32917454	32917455	rs2111504	T	A	10	4	NA	NA	Kawasaki disease
chr19	33364627	33364628	rs8101881	C	T	6	7	NA	NA	Smoking behavior
chr19	33532299	33532300	rs10411210	C	T	399	264	1.58E-07	4.19E-06	Height
chr19	41224203	41224204	rs28493229	G	C	9	7	NA	NA	Alzheimers disease, Alzheimers-AB1-42, HDL cholesterol, LDL cholesterol
chr19	41310570	41310571	rs3733829	A	G	4	6	NA	NA	AB1-42, Brain imaging, C-reactive protein, Cardiovascular disease risk factors, Cholesterol, Longevity
chr19	41937094	41937095	rs17318596	G	A	32	33	1.00E+00	1.00E+00	Alzheimers-AB1-42, p-tau181p
chr19	45395265	45395266	rs157580	G	A	18	7	4.33E-02	1.78E-01	AB1-42, Cholesterol, Triglycerides
chr19	45395618	45395619	rs2075650	A	G	4	6	NA	NA	AB1-42, Alzheimers-disease, Alzheimers-AB1-42, Alzheimers-p-tau181p
chr19	45414450	45414451	rs439401	T	C	425	405	4.88E-01	8.06E-01	AB1-42, Brain imaging, C-reactive protein, Cardiovascular disease risk factors, Cholesterol, Longevity
chr19	47207653	47207654	rs11083846	G	A	5	5	NA	NA	Chronic lymphocytic leukemia
chr19	47268372	47268373	rs11668878	G	T	5	9	NA	NA	Chronic lymphocytic leukemia
chr19	47661492	47661493	rs307896	G	A	53	58	7.04E-01	9.29E-01	Multiple sclerosis
chr19	49208864	49208865	rs504963	G	A	6	16	5.25E-02	2.03E-01	Crohns disease
chr19	49214273	49214274	rs281379	G	A	6	8	NA	NA	Crohns disease

chrom	chromStart	chromEnd	dbSNP_ID	ref_allele	alt_allele	ref_count	alt_count	P-value	q-value	GWAS_traits
chr19	49228271	49228272	rs2287921	T	C	789	843	1.81E-01	4.71E-01	Bipolar disorder, Retinal vascular caliber
chr19	50000008	50000009	rs2280401	G	A	4	6	NA	NA	Albumin, Protein-total
chr19	55819844	55819845	rs1172822	C	T	12	8	NA	NA	Menopause
chr2	634904	634905	rs6548238	T	C	20	25	5.51E-01	8.37E-01	BMI
chr2	1925992	1925993	rs2241685	C	T	4	8	NA	NA	ADHD
chr2	11220561	11220562	rs6739054	A	G	38	51	2.03E-01	5.03E-01	Cognitive_performance-PC1
chr2	20261684	20261685	rs7565124	G	A	10	11	1.00E+00	1.00E+00	Major depressive disorder
chr2	25301754	25301755	rs478222	A	T	38	46	4.45E-01	7.69E-01	Type 1 diabetes
chr2	25887557	25887558	rs11684202	A	G	24	22	8.83E-01	1.00E+00	LDL cholesterol
chr2	27851917	27851918	rs3749147	G	A	1003	973	5.00E-01	8.11E-01	Triglycerides waist circumference
chr2	28646800	28646801	rs6547853	G	A	6	5	NA	NA	Type 1 diabetes
chr2	33701889	33701890	rs13385731	T	C	83	141	1.29E-04	1.51E-03	Systemic lupus erythematosus
chr2	34480074	34480075	rs6733379	G	T	10	12	8.32E-01	1.00E+00	ADHD
chr2	36673554	36673555	rs7562790	T	G	30	25	5.90E-01	8.50E-01	QRS duration
chr2	37750979	37750980	rs4352210	A	G	175	148	1.48E-01	4.15E-01	RR interval
chr2	37940541	37940542	rs4670766	T	C	76	122	1.32E-03	1.12E-02	Alzheimers Total ventricular volume
chr2	42276920	42276921	rs4952590	C	T	48	41	5.25E-01	8.32E-01	Atopy
chr2	42981238	42981239	rs930421	A	G	173	225	1.05E-02	5.95E-02	ADHD
chr2	43359060	43359061	rs12466022	C	A	67	51	1.67E-01	4.49E-01	Multiple sclerosis
chr2	43540124	43540125	rs7590268	T	G	9	15	3.07E-01	6.21E-01	Nonsyndromic cleft lip with or without cleft palate
chr2	43553948	43553949	rs1465618	T	C	11	16	4.42E-01	7.66E-01	Prostate cancer
chr2	43687878	43687879	rs17030845	C	T	21	65	2.15E-06	4.19E-05	Platelet count
chr2	47229838	47229839	rs17540621	G	C	23	10	3.51E-02	1.56E-01	Response to statin therapy-chol sum
chr2	56096891	56096892	rs3791679	A	G	47	136	3.15E-11	2.05E-09	Height
chr2	60762501	60762502	rs2556378	T	G	36	29	4.57E-01	7.72E-01	ADHD
chr2	61066665	61066666	rs1432295	G	A	44	34	3.08E-01	6.21E-01	Hodgkins lymphoma
chr2	61186828	61186829	rs13003464	A	G	10	9	NA	NA	Celiac disease, Ulcerative colitis
chr2	65608908	65608909	rs1876518	C	T	3	8	NA	NA	Rheumatoid arthritis celiac disease
chr2	85699749	85699750	rs7577642	C	T	14	7	1.89E-01	4.82E-01	IL6R
chr2	87044315	87044316	rs6547705	G	A	4	11	NA	NA	Progressive supranuclear palsy
chr2	88924621	88924622	rs11684404	T	C	3	7	NA	NA	Height
chr2	97413487	97413488	rs2314398	C	G	60	47	2.46E-01	5.54E-01	Bipolar disorder
chr2	99239930	99239931	rs12618769	C	T	3	8	NA	NA	Bipolar disorder
chr2	99382891	99382892	rs7578035	T	G	148	50	1.90E-12	1.39E-10	Bipolar disorder
chr2	100825366	100825367	rs9653442	C	T	6	4	NA	NA	Type 1 diabetes
chr2	102953616	102953617	rs3771180	G	T	100	138	1.63E-02	8.49E-02	Asthma
chr2	105837597	105837598	rs6712932	T	C	3	8	NA	NA	Type 2 diabetes
chr2	113841029	113841030	rs6734238	A	G	14	11	6.90E-01	9.20E-01	C-reactive protein
chr2	125281909	125281910	rs17727261	C	T	3	7	NA	NA	Response to antipsychotic therapy risperidone
chr2	134005568	134005569	rs10496702	G	A	13	6	NA	NA	Bipolar disorder and schizophrenia
chr2	137015990	137015991	rs4452212	G	A	6	5	NA	NA	Telomere length
chr2	158780203	158780204	rs10183640	G	A	5	6	NA	NA	Sudden cardiac arrest
chr2	163124050	163124051	rs1990760	C	T	7	4	NA	NA	Immunoglobulin A, Type 1 diabetes, Type 1 diabetes autoantibodies
chr2	163260690	163260691	rs17716942	T	C	15	13	8.51E-01	1.00E+00	Psoriasis
chr2	165577163	165577164	rs10221833	G	C	17	17	1.00E+00	1.00E+00	Response to statin therapy-Triglyceride sum
chr2	169041385	169041386	rs6749447	T	G	8	25	4.55E-03	3.20E-02	Blood pressure
chr2	176991778	176991779	rs2592394	A	G	62	87	4.89E-02	1.96E-01	Serum magnesium levels
chr2	177042632	177042633	rs2072590	A	C	72	66	6.71E-01	9.10E-01	Ovarian cancer
chr2	177909594	177909595	rs1529093	T	C	7	22	8.13E-03	4.95E-02	NAFLDH
chr2	180566677	180566678	rs16866933	G	A	18	9	1.22E-01	3.66E-01	Sudden cardiac arrest
chr2	182323664	182323665	rs12988934	C	T	12	14	8.45E-01	1.00E+00	Monocyte count
chr2	188343496	188343497	rs7586970	T	C	12	13	1.00E+00	1.00E+00	Coronary heart disease
chr2	191538561	191538562	rs10931468	C	A	4	17	7.20E-03	4.52E-02	Primary biliary cirrhosis
chr2	211060049	211060050	rs2286963	T	G	45	41	7.47E-01	9.50E-01	C9 C10-2
chr2	218729864	218729865	rs3791950	C	A	28	15	6.60E-02	2.35E-01	Height

chrom	chromStart	chromEnd	dbSNP_ID	ref_allele	alt_allele	ref_count	alt_count	P-value	q-value	GWAS_traits
chr2	219010145	219010146	rs11676348	C	T	35	21	8.14E-02	2.78E-01	Ulcerative colitis
chr2	219756382	219756383	rs7349332	C	T	7	6	NA	NA	Hair curl, Hair morphology
chr2	221720087	221720088	rs10932886	G	A	12	6	NA	NA	Brain imaging
chr2	223917982	223917983	rs12621643	T	G	53	38	1.42E-01	4.08E-01	Acute lymphoblastic leukemia childhood
chr2	227773465	227773466	rs3820928	T	C	19	21	8.75E-01	1.00E+00	FEV1
chr2	229969797	229969798	rs6436839	A	G	26	37	2.07E-01	5.11E-01	Quetiapine -Schizophrenia neurocognition
chr2	231091222	231091223	rs13397985	T	G	25	31	5.04E-01	8.11E-01	Chronic lymphocytic leukemia
chr2	234673308	234673309	rs4148325	C	T	9	6	NA	NA	Bilirubin
chr2	235210726	235210727	rs12151790	G	A	18	11	2.65E-01	5.75E-01	Osteoporosis
chr2	235900170	235900171	rs11676855	T	C	14	6	NA	NA	Dialysis-related mortality
chr2	238270893	238270894	rs10202497	C	A	16	22	4.18E-01	7.46E-01	Aging traits-age free from disease
chr2	239306267	239306268	rs3739070	A	C	13	3	NA	NA	Iris characteristics
chr2	240418659	240418660	rs6743931	G	A	5	7	NA	NA	Response to antipsychotic therapy extrapyramidal side effects
chr20	2654924	2654925	rs1810636	A	C	77	102	7.25E-02	2.54E-01	Longevity
chr20	3776174	3776175	rs3761218	C	T	79	79	1.00E+00	1.00E+00	Bipolar disorder
chr20	5037773	5037774	rs261360	G	A	326	251	1.79E-03	1.46E-02	Hair morphology
chr20	6035227	6035228	rs2326679	C	T	14	18	5.97E-01	8.50E-01	Menopause
chr20	7180055	7180056	rs6085920	T	A	4	7	NA	NA	Serum urate
chr20	7552503	7552504	rs6108011	C	T	38	27	2.15E-01	5.15E-01	Red blood cell count
chr20	8114703	8114704	rs6118083	A	G	115	92	1.26E-01	3.76E-01	Colorword
chr20	10969029	10969030	rs1327235	A	G	6	14	NA	NA	Blood pressure, Diastolic blood pressure, Systolic blood pressure
chr20	31950844	31950845	rs291671	G	A	5	5	NA	NA	Hair color-red hair
chr20	32738611	32738612	rs1015362	C	T	77	57	1.00E-01	3.20E-01	Burning and freckling, Freckles, Hair color-red hair
chr20	33907160	33907161	rs6060369	T	C	314	207	2.76E-06	5.04E-05	Height
chr20	33914207	33914208	rs6060373	A	G	48	62	2.15E-01	5.15E-01	Height
chr20	33975180	33975181	rs6088813	C	A	5	5	NA	NA	Height
chr20	43065027	43065028	rs6017342	A	C	4	6	NA	NA	Ulcerative colitis
chr20	44740195	44740196	rs6074022	C	T	33	54	3.14E-02	1.44E-01	Multiple sclerosis
chr20	44747946	44747947	rs4810485	T	G	29	47	5.05E-02	1.98E-01	Rheumatoid arthritis
chr20	48522329	48522330	rs495337	G	A	40	11	5.70E-05	7.24E-04	Psoriasis
chr20	48554976	48554977	rs2235617	C	G	27	31	6.94E-01	9.23E-01	Psoriasis
chr20	49375241	49375242	rs6122972	A	G	76	59	1.68E-01	4.49E-01	Bipolar disorder
chr20	52791517	52791518	rs2248359	C	T	8	5	NA	NA	Multiple sclerosis
chr20	53267110	53267111	rs2023454	C	G	12	33	2.46E-03	1.94E-02	Functional MRI
chr20	56044183	56044184	rs6070116	A	G	3	8	NA	NA	Response to statin therapy-chol sum
chr20	59268709	59268710	rs6027755	G	A	17	23	4.30E-01	7.60E-01	NAFLDH
chr20	62318219	62318220	rs4809324	T	C	167	149	3.39E-01	6.58E-01	Glioma-high-grade
chr20	62327581	62327582	rs2297441	G	A	3	7	NA	NA	Ulcerative colitis
chr21	16817050	16817051	rs1297265	A	G	22	6	3.72E-03	2.71E-02	Ulcerative colitis
chr21	17828290	17828291	rs2823819	A	G	21	22	1.00E+00	1.00E+00	ADHD
chr21	20536648	20536649	rs2825388	T	A	6	5	NA	NA	ADHD
chr21	28665346	28665347	rs2830840	G	C	13	17	5.85E-01	8.50E-01	Response to citalopram treatment
chr21	28744355	28744356	rs1452093	A	G	95	110	3.28E-01	6.43E-01	Dialysis-related mortality
chr21	31050816	31050817	rs363512	G	A	8	4	NA	NA	ADHD-Hyperactive-impulsive symptoms
chr21	33285299	33285300	rs2833556	G	A	9	4	NA	NA	Olanzapine Schizophrenia neurocognition
chr21	36416331	36416332	rs9983044	C	G	17	10	2.48E-01	5.54E-01	Serum polyunsaturated fatty acids
chr21	38740823	38740824	rs12483205	A	G	94	108	3.60E-01	6.79E-01	HIV-1 replication
chr21	40173527	40173528	rs1209950	C	T	4	6	NA	NA	Lung cancer-non-small cell
chr21	40465177	40465178	rs2242944	G	A	6	5	NA	NA	Ankylosing spondylitis
chr21	40469519	40469520	rs378108	A	G	27	18	2.33E-01	5.41E-01	Ankylosing spondylitis
chr21	43841327	43841328	rs3788013	C	A	70	42	1.04E-02	5.95E-02	Type 1 diabetes autoantibodies
chr21	44448717	44448718	rs2839627	C	T	12	19	2.81E-01	5.92E-01	Information processing speed speed factor
chr21	45615022	45615023	rs2838519	G	A	120	120	1.00E+00	1.00E+00	Ulcerative colitis
chr22	22307518	22307519	rs412050	G	C	30	40	2.82E-01	5.92E-01	ADHD

chrom	chromStart	chromEnd	dbSNP_ID	ref_allele	alt_allele	ref_count	alt_count	P-value	q-value	GWAS_traits
chr22	24999103	24999104	rs2073398	C	G	62	18	8.14E-07	1.70E-05	Gamma glutamyl transferase
chr22	27042105	27042106	rs744016	C	T	4	6	NA	NA	RR interval
chr22	28090206	28090207	rs5762311	G	A	3	10	NA	NA	Sudden cardiac arrest
chr22	29130011	29130012	rs738722	T	C	21	17	6.27E-01	8.72E-01	Esophageal cancer
chr22	29451670	29451671	rs4823006	A	G	120	105	3.51E-01	6.69E-01	Waist-hip ratio
chr22	30581721	30581722	rs5753037	C	T	9	3	NA	NA	Type 1 diabetes
chr22	30592068	30592069	rs2412980	T	C	351	311	1.20E-01	3.66E-01	Dialysis-related mortality
chr22	30592486	30592487	rs713875	C	G	1764	1309	0.00E+00	0.00E+00	Crohns disease
chr22	31645758	31645759	rs2106294	C	T	43	61	9.50E-02	3.17E-01	Diabetic nephropathy
chr22	37310045	37310046	rs2075726	G	A	28	24	6.78E-01	9.10E-01	Ankylosing spondylitis
chr22	37469590	37469591	rs4820268	G	A	6	4	NA	NA	Iron, Mean corpuscular hemoglobin, Mean corpuscular volume
chr22	37551606	37551607	rs743777	A	G	59	58	1.00E+00	1.00E+00	Rheumatoid arthritis, Type 1 diabetes autoantibodies
chr22	37581484	37581485	rs229527	C	A	7	10	NA	NA	Vitiligo
chr22	38544297	38544298	rs2284063	A	G	17	32	4.44E-02	1.81E-01	Cutaneous nevi, Melanoma
chr22	38569005	38569006	rs738322	A	G	273	553	0.00E+00	3.90E-14	Cutaneous nevi
chr22	40652872	40652873	rs12484776	A	G	12	16	5.72E-01	8.50E-01	Uterine fibroids
chr22	43500211	43500212	rs5759167	G	T	375	243	1.10E-07	3.20E-06	Prostate cancer
chr22	50966913	50966914	rs470119	T	C	8	12	NA	NA	Mean corpuscular hemoglobin
chr22	50971751	50971752	rs131794	A	C	4	7	NA	NA	Mean corpuscular volume
chr3	4437357	4437358	rs794185	T	C	80	76	8.10E-01	1.00E+00	Multiple sclerosis-Brain Glutamate Concentrations
chr3	6903296	6903297	rs3749380	C	T	33	31	9.01E-01	1.00E+00	Panic disorder
chr3	7529554	7529555	rs9870680	C	T	5	8	NA	NA	Major depressive disorder broad
chr3	11040438	11040439	rs9990174	G	T	3	7	NA	NA	Conduct disorder interaction
chr3	15440555	15440556	rs6442522	C	T	7	3	NA	NA	Serum urate
chr3	19375790	19375791	rs2053506	G	A	5	5	NA	NA	Hip geometry
chr3	28705763	28705764	rs7617877	A	G	4	8	NA	NA	Parkinsons disease
chr3	29575462	29575463	rs1530057	G	T	36	35	1.00E+00	1.00E+00	Lung cancer
chr3	33015468	33015469	rs13314993	G	T	6	4	NA	NA	Celiac disease
chr3	38657898	38657899	rs11710077	A	T	20	18	8.71E-01	1.00E+00	QRS duration
chr3	38767314	38767315	rs6801957	T	C	96	159	9.59E-05	1.14E-03	QRS duration
chr3	39555579	39555580	rs864643	A	G	25	23	8.85E-01	1.00E+00	ADHD
chr3	40096617	40096618	rs6599077	G	A	122	114	6.49E-01	8.94E-01	Sleep duration
chr3	45731450	45731451	rs2742417	C	T	56	52	7.73E-01	9.71E-01	Response to antidepressants-bupropion
chr3	46339811	46339812	rs9990343	A	G	13	17	5.85E-01	8.50E-01	Brain structure
chr3	46428189	46428190	rs6441975	A	C	5	6	NA	NA	HIV progression
chr3	49210731	49210732	rs7617480	A	C	54	52	9.23E-01	1.00E+00	Menarche
chr3	53118738	53118739	rs2336725	C	T	18	25	3.60E-01	6.79E-01	Height
chr3	55571759	55571760	rs1795648	G	A	11	11	1.00E+00	1.00E+00	Schizophrenia
chr3	56667681	56667682	rs9835332	G	C	11	8	NA	NA	Height
chr3	58370176	58370177	rs6445975	G	T	162	116	6.85E-03	4.35E-02	Systemic lupus erythematosus
chr3	61794053	61794054	rs652889	C	A	30	22	3.32E-01	6.46E-01	QT interval
chr3	69252898	69252899	rs6806528	C	T	15	26	1.17E-01	3.62E-01	Celiac disease
chr3	72437412	72437413	rs9863706	C	T	7	5	NA	NA	Height
chr3	87173323	87173324	rs17181170	G	A	4	10	NA	NA	Prostate cancer
chr3	102203044	102203045	rs2063640	C	A	16	14	8.56E-01	1.00E+00	Type 2 diabetes
chr3	105586713	105586714	rs9657904	T	C	25	20	5.51E-01	8.37E-01	Multiple sclerosis
chr3	109927375	109927376	rs13323983	A	G	11	9	NA	NA	Neutrophil count
chr3	119219933	119219934	rs2293370	G	A	19	12	2.81E-01	5.92E-01	Primary biliary cirrhosis
chr3	121660663	121660664	rs4285028	A	C	10	9	NA	NA	Multiple sclerosis
chr3	123094450	123094451	rs2877716	T	C	11	19	2.00E-01	4.98E-01	Two-hour glucose challenge
chr3	141102832	141102833	rs6763931	G	A	162	148	4.60E-01	7.73E-01	Height, Prostate cancer
chr3	141105569	141105570	rs724016	A	G	197	213	4.59E-01	7.72E-01	Height
chr3	141807136	141807137	rs347685	C	A	7	3	NA	NA	Creatinine
chr3	143056466	143056467	rs7632299	G	A	59	58	1.00E+00	1.00E+00	NAFLDH
chr3	156854741	156854742	rs13064954	G	A	49	46	8.38E-01	1.00E+00	Diabetic retinopathy
chr3	159665049	159665050	rs17810546	A	G	91	83	5.96E-01	8.50E-01	Celiac disease
chr3	159728877	159728878	rs6441286	T	G	5	7	NA	NA	Primary biliary cirrhosis
chr3	159745862	159745863	rs485499	T	C	189	168	2.90E-01	6.02E-01	Primary biliary cirrhosis
chr3	169481270	169481271	rs12696304	C	G	12	10	8.32E-01	1.00E+00	Telomere length
chr3	169492100	169492101	rs10936599	C	T	11	13	8.39E-01	1.00E+00	Celiac disease, Colorectal cancer, Multiple sclerosis

chrom	chromStart	chromEnd	dbSNP_ID	ref_allele	alt_allele	ref_count	alt_count	P-value	q-value	GWAS_traits
chr3	170717520	170717521	rs11920090	T	A	6	28	1.95E-04	2.11E-03	Fasting plasma glucose, Insulin resistance
chr3	170971290	170971291	rs2088885	C	A	11	16	4.42E-01	7.66E-01	Brain imaging in schizophrenia interaction
chr3	177596988	177596989	rs7612209	G	A	18	11	2.65E-01	5.75E-01	Cognitive performance-IED
chr3	180551213	180551214	rs1879248	C	T	7	10	NA	NA	Schizophrenia
chr3	182821274	182821275	rs11711441	G	A	12	6	NA	NA	Parkinsons disease
chr3	182869817	182869818	rs683395	G	A	21	8	2.41E-02	1.19E-01	Bipolar disorder
chr3	184514612	184514613	rs4686760	G	A	13	16	7.11E-01	9.35E-01	Van Willebrand factor antibodies
chr3	185531660	185531661	rs1374910	C	T	18	32	6.49E-02	2.35E-01	Type 2 diabetes
chr3	185629567	185629568	rs2002675	A	G	12	11	1.00E+00	1.00E+00	Menarche
chr3	186650789	186650790	rs10937275	A	G	35	30	6.20E-01	8.67E-01	Flucloxacillin-induced liver injury
chr3	188112553	188112554	rs1464510	C	A	15	9	3.07E-01	6.21E-01	Celiac disease, Vitiligo
chr3	191000807	191000808	rs10937470	C	T	76	110	1.53E-02	8.15E-02	Alzheimers Total ventricular volume
chr3	194327097	194327098	rs789852	T	C	6	5	NA	NA	QT interval
chr3	194858373	194858374	rs2131877	G	A	18	17	1.00E+00	1.00E+00	Lung cancer-non-small cell
chr3	195800546	195800547	rs9859260	C	T	7	6	NA	NA	Mean corpuscular volume
chr4	964358	964359	rs11248060	C	T	9	12	6.64E-01	9.08E-01	Parkinsons disease
chr4	6891518	6891519	rs11734132	G	C	4	6	NA	NA	Mean platelet volume
chr4	10056375	10056376	rs7671266	T	C	21	37	4.79E-02	1.93E-01	Cardiovascular disease risk factors
chr4	15737347	15737348	rs4698412	G	A	33	11	1.26E-03	1.11E-02	Parkinsons disease
chr4	25810095	25810096	rs959903	G	A	4	7	NA	NA	NAFLDH
chr4	26108196	26108197	rs874040	G	C	7	20	1.92E-02	9.81E-02	Rheumatoid arthritis
chr4	27744717	27744718	rs4692256	C	T	30	28	8.96E-01	1.00E+00	Brain imaging
chr4	40303632	40303633	rs6832151	G	T	97	107	5.29E-01	8.32E-01	Graves disease
chr4	54799244	54799245	rs871606	T	C	141	180	3.38E-02	1.52E-01	Blood pressure
chr4	55092625	55092626	rs2114039	T	C	67	59	5.33E-01	8.32E-01	Corneal curvature
chr4	68447248	68447249	rs2242330	A	G	7	8	NA	NA	Parkinsons disease
chr4	73515132	73515133	rs7697556	T	C	15	3	NA	NA	Height
chr4	74977836	74977837	rs1371799	T	C	386	360	3.41E-01	6.60E-01	WBC count
chr4	77198985	77198986	rs6812193	C	T	11	10	1.00E+00	1.00E+00	Parkinsons disease
chr4	77368846	77368847	rs17319721	G	A	97	117	1.94E-01	4.88E-01	Creatinine
chr4	77412139	77412140	rs13146355	G	A	5	11	NA	NA	Serum magnesium levels
chr4	84374479	84374480	rs1494961	C	T	16	10	3.27E-01	6.43E-01	oral pharyngeal laryngeal cancer esophageal cancer
chr4	88755827	88755828	rs7698623	T	C	5	8	NA	NA	Stroke
chr4	90678540	90678541	rs2736990	G	A	4	6	NA	NA	Parkinsons disease
chr4	95514608	95514609	rs12500426	A	C	3	8	NA	NA	Prostate cancer
chr4	95562876	95562877	rs17021918	C	T	8	4	NA	NA	Prostate cancer
chr4	100274285	100274286	rs1789924	C	T	243	171	4.70E-04	4.65E-03	oral pharyngeal laryngeal cancer esophageal cancer
chr4	103551602	103551603	rs7665090	A	G	8	4	NA	NA	Primary biliary cirrhosis
chr4	103578636	103578637	rs228614	G	A	12	7	NA	NA	Multiple sclerosis
chr4	122665513	122665514	rs7659604	C	T	28	10	5.10E-03	3.54E-02	Type 2 diabetes
chr4	123377979	123377980	rs2069762	A	C	10	11	1.00E+00	1.00E+00	Type 1 diabetes autoantibodies
chr4	123523874	123523875	rs7682241	G	T	178	190	5.66E-01	8.48E-01	Alopecia areata
chr4	141387437	141387438	rs1594468	C	T	13	9	5.23E-01	8.32E-01	Bilirubin
chr4	142142728	142142729	rs17007017	A	G	222	197	2.41E-01	5.54E-01	Conduct disorder case status
chr4	142709722	142709723	rs17007695	T	C	3	8	NA	NA	Response to treatment for acute lymphoblastic leukemia
chr4	145485737	145485738	rs1980057	C	T	40	51	2.94E-01	6.10E-01	FEV1/FVC
chr4	145486388	145486389	rs13118928	A	G	5	5	NA	NA	COPD
chr4	146821409	146821410	rs4835265	C	A	4	23	3.11E-04	3.18E-03	Gamma glutamyl transferase
chr4	148974601	148974602	rs6845865	T	C	32	35	8.07E-01	9.99E-01	QT interval
chr4	154153999	154154000	rs12644284	A	G	31	32	1.00E+00	1.00E+00	Multiple sclerosis
chr4	154703595	154703596	rs17030434	T	C	27	25	8.90E-01	1.00E+00	P wave duration
chr4	155483913	155483914	rs1800788	C	T	19	18	1.00E+00	1.00E+00	Fibrinogen
chr4	187678865	187678866	rs925642	C	T	30	22	3.32E-01	6.46E-01	Obesity
chr5	1113243	1113244	rs4580814	G	T	178	167	5.90E-01	8.50E-01	Mean corpuscular hemoglobin
chr5	1315659	1315660	rs4975616	G	A	27	18	2.33E-01	5.41E-01	Lung cancer
chr5	4029788	4029789	rs11748327	C	T	3	22	1.57E-04	1.79E-03	Myocardial infarction
chr5	32804527	32804528	rs1173766	T	C	4	8	NA	NA	Diastolic blood pressure, Systolic blood pressure
chr5	32815027	32815028	rs1173771	A	G	168	207	4.96E-02	1.97E-01	Blood pressure, Diastolic blood

chrom	chromStart	chromEnd	dbSNP_ID	ref_allele	alt_allele	ref_count	alt_count	P-value	q-value	GWAS_traits
chr5	35803576	35803577	rs931555	C	T	77	56	8.25E-02	2.80E-01	pressure, Systolic blood pressure
chr5	40401508	40401509	rs17234657	T	G	52	66	2.31E-01	5.41E-01	Multiple sclerosis
chr5	40410583	40410584	rs11742570	T	C	546	611	5.60E-02	2.11E-01	Crohns disease
chr5	40410934	40410935	rs6451493	G	T	181	159	2.55E-01	5.66E-01	Crohns disease
chr5	40437947	40437948	rs9292777	C	T	310	132	0.00E+00	2.00E-15	Ulcerative colitis
chr5	40791883	40791884	rs13361707	C	T	81	81	1.00E+00	1.00E+00	Crohns disease
chr5	44365544	44365545	rs2121875	C	A	16	14	8.56E-01	1.00E+00	Gastric cancer
chr5	55438579	55438580	rs6859219	C	A	9	23	2.01E-02	1.02E-01	Prostate cancer
chr5	55854152	55854153	rs6867983	C	T	11	13	8.39E-01	1.00E+00	Rheumatoid arthritis
chr5	56031883	56031884	rs889312	C	A	50	41	4.02E-01	7.32E-01	Triglycerides
chr5	56031883	56031884	rs889312	C	A	58	63	7.16E-01	9.36E-01	Breast cancer
chr5	65739439	65739439	rs10514995	T	C	3	10	NA	NA	RR interval
chr5	77291939	77291940	rs12655917	G	A	31	31	1.00E+00	1.00E+00	Insulin resistance
chr5	77630853	77630854	rs6881634	G	A	4	7	NA	NA	Hippocampal atrophy
chr5	78272941	78272942	rs2052550	T	C	122	98	1.21E-01	3.66E-01	Ferritin
chr5	88088438	88088439	rs770189	G	C	25	6	8.78E-04	7.97E-03	Tonometry
chr5	102596719	102596720	rs26232	C	T	34	44	3.08E-01	6.21E-01	Rheumatoid arthritis
chr5	106724587	106724588	rs252817	C	T	NA	NA	NA	NA	Bi polar disorder and schizophrenia
chr5	109496959	109496960	rs4460176	T	G	6	4	NA	NA	Plasminogen activator inhibitor 1
chr5	110401871	110401872	rs1837253	T	C	6	5	NA	NA	Asthma
chr5	110405674	110405675	rs3806932	A	G	66	157	9.87E-10	4.80E-08	Eosinophilic esophagitis pediatric
chr5	112242967	112242968	rs469727	C	T	41	42	1.00E+00	1.00E+00	ADHD
chr5	112996653	112996654	rs17135859	T	C	41	33	4.16E-01	7.46E-01	F-cell distribution
chr5	124332102	124332103	rs4836133	C	A	80	24	3.22E-08	1.11E-06	BMI
chr5	127640860	127640861	rs27855	A	G	6	5	NA	NA	Height
chr5	131770804	131770805	rs2188962	C	T	8	4	NA	NA	Crohns disease
chr5	131801725	131801726	rs2522056	G	A	6	4	NA	NA	Fibrinogen
chr5	131819920	131819921	rs2070729	C	A	6	6	NA	NA	Platelet count
chr5	131862976	131862977	rs4143832	T	G	11	20	1.50E-01	4.18E-01	Plasma eosinophil count
chr5	134467699	134467700	rs477687	G	A	24	14	1.43E-01	4.09E-01	HIV progression
chr5	141522999	141523000	rs1062158	C	T	11	9	NA	NA	Multiple sclerosis
chr5	147845814	147845815	rs3995090	A	C	9	21	4.28E-02	1.78E-01	FEV1
chr5	150458145	150458146	rs10036748	C	T	10	3	NA	NA	Systemic lupus erythematosus
chr5	152979553	152979554	rs12658202	C	A	3	7	NA	NA	Height
chr5	158759899	158759900	rs2546890	A	G	5	5	NA	NA	Multiple sclerosis, Psoriasis
chr5	159879977	159879978	rs2431697	T	C	21	19	8.75E-01	1.00E+00	Systemic lupus erythematosus
chr5	166389492	166389493	rs17404956	A	G	15	8	2.10E-01	5.13E-01	Diabetic retinopathy
chr5	168256239	168256240	rs4282339	G	A	18	14	5.97E-01	8.50E-01	Height
chr5	169074055	169074056	rs169082	C	T	19	13	3.77E-01	6.99E-01	Leptin
chr5	169950393	169950394	rs11957313	G	A	71	86	2.64E-01	5.75E-01	Normalized brain volume
chr5	171076226	171076227	rs254893	G	A	20	31	1.61E-01	4.39E-01	Diastolic blood pressure
chr5	176784511	176784512	rs4075958	G	A	7	9	NA	NA	Multiple sclerosis
chr5	176817635	176817636	rs6420094	A	G	341	328	6.15E-01	8.65E-01	Creatinine
chr5	180595137	180595138	rs1279750	C	T	15	11	5.57E-01	8.39E-01	Platelet count
chr6	411063	411064	rs872071	A	G	5	16	2.66E-02	1.26E-01	Chronic lymphocytic leukemia
chr6	417726	417727	rs9378805	A	C	7	4	NA	NA	Chronic lymphocytic leukemia
chr6	466032	466033	rs1540771	C	T	6	7	NA	NA	Freckles
chr6	1535997	1535998	rs11242704	A	G	11	10	1.00E+00	1.00E+00	Response to hepatitis C treatment
chr6	5260935	5260936	rs2224391	A	C	297	309	6.26E-01	8.72E-01	Height
chr6	10163967	10163968	rs13195786	A	G	58	66	5.30E-01	8.32E-01	Serum calcium
chr6	10969140	10969141	rs4713103	G	T	72	136	1.08E-05	1.80E-04	Serum polyunsaturated fatty acids
chr6	13332466	13332467	rs499818	G	A	24	11	4.10E-02	1.73E-01	Major CVD
chr6	16742032	16742033	rs697739	C	T	39	26	1.36E-01	3.93E-01	Amyotrophic lateral sclerosis-age of onset
chr6	20652716	20652717	rs9295474	C	G	16	21	5.11E-01	8.20E-01	Type 2 diabetes
chr6	20657563	20657564	rs4712523	A	G	5	6	NA	NA	Type 2 diabetes
chr6	20661249	20661250	rs7754840	G	C	8	5	NA	NA	Type 2 diabetes
chr6	21384612	21384613	rs9466056	A	G	155	208	6.27E-03	4.21E-02	Bone mineral density-femoral neck
chr6	22017737	22017738	rs1928168	T	C	83	46	1.43E-03	1.19E-02	FEV1/FVC
chr6	25821769	25821770	rs17342717	C	T	9	12	6.64E-01	9.08E-01	Ferritin, Mean corpuscular hemoglobin
chr6	25842950	25842951	rs1408272	T	G	15	15	1.00E+00	1.00E+00	Mean corpuscular hemoglobin, Transferrin receptor

chrom	chromStart	chromEnd	dbSNP_ID	ref_allele	alt_allele	ref_count	alt_count	P-value	q-value	GWAS_traits
chr6	26233386	26233387	rs10946808	A	G	180	194	5.02E-01	8.11E-01	Height
chr6	26500562	26500563	rs13194984	G	T	19	10	1.36E-01	3.93E-01	Transferrin saturation
chr6	28322295	28322296	rs6903823	A	G	25	24	1.00E+00	1.00E+00	FEV1/FVC
chr6	31903803	31903804	rs9332739	G	C	7	7	NA	NA	Age-related macular degeneration
chr6	32050757	32050758	rs1150754	C	T	80	106	6.65E-02	2.35E-01	Systemic lupus erythematosus
chr6	32223257	32223258	rs3130320	T	C	208	330	1.44E-07	4.01E-06	Systemic lupus erythematosus
chr6	32337629	32337630	rs2273017	G	A	571	508	5.51E-02	2.10E-01	Graves disease
chr6	34302868	34302869	rs206936	A	G	5	6	NA	NA	BMI
chr6	36623378	36623379	rs9470361	G	A	58	66	5.30E-01	8.32E-01	QRS duration
chr6	41623032	41623033	rs2143678	G	T	6	5	NA	NA	Major CVD
chr6	41650735	41650736	rs2842643	C	T	6	5	NA	NA	ADHD
chr6	41905274	41905275	rs3218097	G	A	6	4	NA	NA	Red blood cell count
chr6	43795967	43795968	rs943072	G	T	19	19	1.00E+00	1.00E+00	Ulcerative colitis
chr6	43806608	43806609	rs881858	G	A	376	413	1.88E-01	4.82E-01	Creatinine
chr6	44837355	44837356	rs3799977	T	G	7	5	NA	NA	ADHD
chr6	53123117	53123118	rs735860	T	C	13	17	5.85E-01	8.50E-01	Glaucoma
chr6	87466491	87466492	rs493187	A	G	4	8	NA	NA	Bipolar disorder and schizophrenia
chr6	90926611	90926612	rs10806425	C	A	23	24	1.00E+00	1.00E+00	Celiac disease
chr6	90996768	90996769	rs12212193	A	G	287	291	8.68E-01	1.00E+00	Multiple sclerosis
chr6	91207350	91207351	rs806276	A	G	17	3	NA	NA	ADHD
chr6	91417751	91417752	rs713155	G	T	8	3	NA	NA	Brain structure
chr6	105400836	105400837	rs314280	A	G	22	24	8.83E-01	1.00E+00	Menarche
chr6	106094056	106094057	rs9386463	A	G	17	15	8.60E-01	1.00E+00	Primary tooth development time to first tooth eruption
chr6	106435268	106435269	rs7746082	G	C	31	31	1.00E+00	1.00E+00	Crohns disease
chr6	106568033	106568034	rs548234	C	T	5	6	NA	NA	Systemic lupus erythematosus
chr6	106899225	106899226	rs1417352	C	T	4	7	NA	NA	Atrial natriuretic peptide
chr6	109268049	109268050	rs2798641	C	T	140	141	1.00E+00	1.00E+00	FEV1/FVC
chr6	109616419	109616420	rs9374080	T	C	8	5	NA	NA	Mean corpuscular volume
chr6	109742014	109742015	rs9487094	G	A	4	27	3.40E-05	4.61E-04	Height
chr6	111913261	111913262	rs33980500	C	T	64	19	7.39E-07	1.60E-05	Psoriatic arthritis, Psoriasis
chr6	117086377	117086378	rs654128	C	A	30	31	1.00E+00	1.00E+00	Telomere length
chr6	127452638	127452639	rs9491696	C	G	27	28	1.00E+00	1.00E+00	Waist-hip ratio
chr6	134214524	134214525	rs12190287	C	G	17	18	1.00E+00	1.00E+00	Coronary heart disease
chr6	135494874	135494875	rs9321490	T	C	34	38	7.24E-01	9.37E-01	Multiple sclerosis
chr6	135739354	135739355	rs1154801	C	A	6	5	NA	NA	Multiple sclerosis
chr6	138199416	138199417	rs610604	G	T	84	56	2.22E-02	1.12E-01	Psoriasis
chr6	141169824	141169825	rs11155133	A	G	15	17	8.60E-01	1.00E+00	Acute lymphoblastic leukemia childhood
chr6	142679571	142679572	rs6570507	G	A	12	8	NA	NA	Height
chr6	142750515	142750516	rs3817928	A	G	46	53	5.47E-01	8.36E-01	FEV1/FVC
chr6	148704953	148704954	rs6930576	G	A	5	4	NA	NA	Diabetic nephropathy
chr6	150913960	150913961	rs9372078	T	A	7	3	NA	NA	Panic disorder
chr6	152068363	152068364	rs1999805	G	A	4	9	NA	NA	Bone mineral density-spine
chr6	157441049	157441050	rs9478751	A	G	20	20	1.00E+00	1.00E+00	Serum urate
chr6	159465976	159465977	rs1738074	T	C	105	107	9.45E-01	1.00E+00	Celiac disease, Multiple sclerosis
chr6	159490435	159490436	rs212388	C	T	4	7	NA	NA	Crohns disease, Crohns disease celiac disease
chr6	160137686	160137687	rs4516970	G	A	42	56	1.89E-01	4.82E-01	Ferritin
chr6	160402704	160402705	rs6917747	G	A	67	43	2.79E-02	1.31E-01	Brain lesion load
chr6	160581373	160581374	rs651164	A	G	4	6	NA	NA	Prostate cancer
chr6	164186676	164186677	rs10945919	A	G	21	20	1.00E+00	1.00E+00	Response to TNF antagonist treatment
chr6	167383074	167383075	rs9355610	G	A	19	18	1.00E+00	1.00E+00	Graves disease
chr6	167532792	167532793	rs3093024	A	G	9	11	NA	NA	Rheumatoid arthritis
chr6	167534289	167534290	rs3093023	G	A	25	19	4.51E-01	7.72E-01	Rheumatoid arthritis
chr6	169958981	169958982	rs3734905	G	A	8	9	NA	NA	HIV progression
chr7	1886534	1886535	rs6952808	G	C	17	25	2.80E-01	5.92E-01	Bipolar disorder and schizophrenia
chr7	2041431	2041432	rs1107592	A	G	18	12	3.62E-01	6.79E-01	Bipolar disorder and schizophrenia
chr7	2789879	2789880	rs798502	A	C	31	13	9.56E-03	5.53E-02	Ulcerative colitis
chr7	7268430	7268431	rs10259085	C	T	11	4	NA	NA	Multiple sclerosis
chr7	17561582	17561583	rs10499504	A	G	13	31	9.56E-03	5.53E-02	Response to antipsychotic

chrom	chromStart	chromEnd	dbSNP_ID	ref_allele	alt_allele	ref_count	alt_count	P-value	q-value	GWAS_traits	
chr7	20647014	20647015	rs10950821	C	G	16	18	8.64E-01	1.00E+00	therapy clozapine-total cholesterol	
chr7	23502973	23502974	rs12534093	T	A	5	9	NA	NA	Response to statin therapy-acylcarnitine	
chr7	25871108	25871109	rs1055144	C	T	6	6	NA	NA	Height	
chr7	25901638	25901639	rs12700667	G	A	89	81	5.91E-01	8.50E-01	Waist-hip ratio	
chr7	27976562	27976563	rs10486567	G	A	15	20	5.00E-01	8.11E-01	Endometriosis	
chr7	28189410	28189411	rs1635852	T	C	16	6	5.25E-02	2.03E-01	Prostate cancer	
chr7	28391141	28391142	rs11765845	A	G	6	4	NA	NA	Height	
chr7	29041189	29041190	rs2252521	T	C	6	4	NA	NA	Diabetic retinopathy	
chr7	31155346	31155347	rs158477	T	C	12	19	2.81E-01	5.92E-01	Cognitive performance-PAL8	
chr7	36610492	36610493	rs6948404	T	C	5	5	NA	NA	Major depressive disorder	
chr7	44229067	44229068	rs1799884	C	T	12	13	1.00E+00	1.00E+00	HIV progression	
chr7	44235667	44235668	rs4607517	G	A	63	51	3.03E-01	6.21E-01	Fasting plasma glucose, HbA1C	
chr7	46753552	46753553	rs700752	C	G	39	44	6.61E-01	9.08E-01	Fasting plasma glucose, Insulin resistance	
chr7	50305862	50305863	rs4917014	T	G	7	8	NA	NA	IGF1, IGFBP3	
chr7	50622711	50622712	rs1451375	C	A	10	10	NA	NA	Systemic lupus erythematosus	
chr7	55159348	55159349	rs11979158	A	G	18	21	7.49E-01	9.50E-01	Malaria	
chr7	65617594	65617595	rs875971	T	C	52	38	1.70E-01	4.52E-01	Glioma	
chr7	72971230	72971231	rs13233571	C	T	43	43	1.00E+00	1.00E+00	Aortic root size	
chr7	72987353	72987354	rs2286276	C	T	24	12	6.52E-02	2.35E-01	C-reactive protein	
chr7	75953296	75953297	rs758944	C	A	6	5	NA	NA	Triglycerides	
chr7	75975585	75975586	rs7779014	C	T	79	86	6.41E-01	8.89E-01	Multiple sclerosis	
chr7	92264409	92264410	rs2282978	T	C	624	775	5.41E-05	7.02E-04	Multiple sclerosis	
chr7	92408369	92408370	rs445	C	T	336	322	5.85E-01	8.50E-01	Height	
chr7	94053999	94054000	rs441051	T	C	27	23	6.72E-01	9.10E-01	Neutrophil count, WBC count	
chr7	97816326	97816327	rs6465657	C	T	198	266	1.84E-03	1.47E-02	Mean FVC	
chr7	100486753	100486754	rs1266989	G	C	45	42	8.30E-01	1.00E+00	Prostate cancer	
chr7	106359215	106359216	rs342275	C	T	278	284	8.00E-01	9.94E-01	RR interval	
chr7	106372218	106372219	rs342293	C	G	176	121	2.00E-01	1.38E-02	Platelet count	
chr7	106763217	106763218	rs13224682	G	A	48	61	2.50E-01	5.58E-01	Mean platelet volume	
chr7	107207694	107207695	rs4730250	A	G	7	3	NA	NA	Osteoarthritis	
chr7	107479518	107479519	rs4730273	C	A	8	5	NA	NA	Ulcerative colitis	
chr7	107580838	107580839	rs2158836	A	G	15	18	7.28E-01	9.39E-01	Ulcerative colitis	
chr7	110047470	110047471	rs6968385	C	T	9	5	NA	NA	ADHD	
chr7	112628372	112628373	rs10229603	T	C	11	13	8.39E-01	1.00E+00	ADHD	
chr7	116162728	116162729	rs4236601	G	A	11	13	8.39E-01	1.00E+00	Glaucoma	
chr7	116186240	116186241	rs3807989	A	G	128	74	1.77E-04	1.99E-03	PR interval	
chr7	131287989	131287990	rs12534221	C	A	27	23	6.72E-01	9.10E-01	1.00E+00	Alzheimers-AB1-42
chr8	9178267	9178268	rs6984305	A	T	7	3	NA	NA	Hypertension	
chr8	10071619	10071620	rs11775334	A	G	5	4	NA	NA	Alkaline phosphatase	
chr8	11359637	11359638	rs1600249	G	T	7	3	NA	NA	Rheumatoid arthritis	
chr8	19819723	19819724	rs328	C	G	18	25	3.60E-01	6.79E-01	Triglycerides	
chr8	19824491	19824492	rs13702	T	C	29	26	7.88E-01	9.85E-01	HDL cholesterol triglycerides	
chr8	19865174	19865175	rs2083637	A	G	12	22	1.21E-01	3.66E-01	HDL cholesterol	
chr8	23082970	23082971	rs13278062	G	T	262	152	7.14E-08	2.19E-06	Age-related macular degeneration	
chr8	23526462	23526463	rs1512268	T	C	11	9	NA	NA	Menopause	
chr8	27452846	27452847	rs17466684	G	A	28	18	1.84E-01	4.75E-01	Prostate cancer	
chr8	27466314	27466315	rs1532278	T	C	202	226	2.66E-01	5.76E-01	Panic disorder	
chr8	32412358	32412359	rs2439312	A	G	543	590	1.63E-01	4.40E-01	Alzheimers disease late onset	
chr8	37686748	37686749	rs6468442	A	G	4	7	NA	NA	Dialysis-related mortality	
chr8	38031344	38031345	rs16887244	A	G	7	6	NA	NA	Schizophrenia	
chr8	38469302	38469303	rs7832232	A	G	6	7	NA	NA	Pancreatic cancer	
chr8	49812200	49812201	rs12155623	A	T	6	4	NA	NA	Sudden cardiac arrest	
chr8	55437523	55437524	rs9298506	A	G	17	20	7.43E-01	9.49E-01	Intracranial aneurysm	
chr8	61003896	61003897	rs6995588	C	T	6	6	NA	NA	Cardiac hypertrophy	
chr8	62409427	62409428	rs903027	G	T	10	4	NA	NA	Alzheimers-Whole-brain volume	
chr8	72263955	72263956	rs2218488	C	T	4	6	NA	NA	Sleepiness	
chr8	95960510	95960511	rs896854	T	C	34	41	4.89E-01	8.06E-01	Type 2 diabetes	
chr8	101919320	101919321	rs931812	C	T	122	333	0.00E+00	0.00E+00	ADHD	
chr8	106581527	106581528	rs6993770	A	T	13	17	5.85E-01	8.50E-01	Platelet count, Vascular endothelial growth factor levels	

chrom	chromStart	chromEnd	dbSNP_ID	ref_allele	alt_allele	ref_count	alt_count	P-value	q-value	GWAS_traits
chr8	116394074	116394075	rs17658378	A	G	3	11	NA	NA	ADHD
chr8	117630682	117630683	rs16892766	A	C	59	26	4.47E-04	4.50E-03	Colorectal cancer
chr8	118827838	118827839	rs11989122	G	A	7	6	NA	NA	Height
chr8	124696866	124696867	rs2294015	C	T	5	6	NA	NA	Alcohol dependence
chr8	126477977	126477978	rs2001945	G	C	37	38	1.00E+00	1.00E+00	Triglycerides
chr8	126540050	126540051	rs1551398	G	A	70	45	2.48E-02	1.20E-01	Crohns disease
chr8	128124915	128124916	rs16901979	C	A	16	15	1.00E+00	1.00E+00	Prostate cancer
chr8	128387851	128387852	rs1562430	T	C	41	24	4.64E-02	1.88E-01	Breast cancer
chr8	128407442	128407443	rs10505477	A	G	53	51	9.22E-01	1.00E+00	Colorectal cancer
chr8	128413304	128413305	rs6983267	G	T	147	103	6.42E-03	4.25E-02	Colorectal cancer, Prostate cancer
chr8	128517572	128517573	rs4242382	A	G	31	13	9.56E-03	5.53E-02	Prostate cancer
chr8	128815028	128815029	rs4410871	T	C	35	25	2.45E-01	5.54E-01	Multiple sclerosis
chr8	129946153	129946154	rs987525	C	A	6	5	NA	NA	Cleft lip, Nonsyndromic cleft lip with or without cleft palate
chr8	130491751	130491752	rs891835	T	G	7	5	NA	NA	Glioma
chr8	130685456	130685457	rs4295627	T	G	10	17	2.48E-01	5.54E-01	Glioma
chr8	135611944	135611945	rs1036819	A	C	6	4	NA	NA	Longevity
chr8	142300314	142300315	rs7827290	T	G	24	33	2.89E-01	6.02E-01	Bipolar disorder
chr8	142359549	142359550	rs7386474	A	G	7	5	NA	NA	Bipolar disorder and schizophrenia
chr9	1056958	1056959	rs17641078	G	C	6	11	NA	NA	ADHD-Hyperactive-impulsive symptoms
chr9	4763175	4763176	rs385893	T	C	80	94	3.24E-01	6.43E-01	Platelet count
chr9	15296033	15296034	rs643531	C	A	126	99	8.28E-02	2.80E-01	HDL cholesterol
chr9	16864520	16864521	rs2153271	C	T	36	32	7.16E-01	9.36E-01	Freckling
chr9	21986846	21986847	rs3731211	T	A	27	19	3.02E-01	6.21E-01	Platelet count
chr9	22098573	22098574	rs4977574	A	G	34	16	1.53E-02	8.15E-02	Coronary heart disease, Myocardial infarction
chr9	22136488	22136489	rs1333051	A	T	54	48	6.21E-01	8.67E-01	Type 2 diabetes
chr9	27536396	27536397	rs2814707	C	T	12	4	NA	NA	Amyotrophic lateral sclerosis
chr9	34710259	34710260	rs2812378	G	A	169	143	1.57E-01	4.32E-01	Rheumatoid arthritis
chr9	71865931	71865932	rs2282335	G	A	8	8	NA	NA	Renal sinus fat
chr9	75764564	75764565	rs3758354	A	C	191	339	1.29E-10	7.34E-09	Schizophrenia bipolar disorder and depression
chr9	79038169	79038170	rs6560517	T	G	3	7	NA	NA	Dialysis-related mortality
chr9	92887230	92887231	rs2081670	C	T	8	5	NA	NA	Macrophage inflammatory factor 1B
chr9	93563535	93563536	rs290986	A	G	22	35	1.11E-01	3.47E-01	Multiple sclerosis
chr9	93975470	93975471	rs773506	G	A	14	9	4.05E-01	7.32E-01	Diabetic nephropathy
chr9	98259702	98259703	rs10512248	T	G	15	17	8.60E-01	1.00E+00	Height
chr9	107594363	107594364	rs2515629	A	G	5	6	NA	NA	HDL cholesterol
chr9	117049890	117049891	rs946053	T	G	70	46	3.23E-02	1.46E-01	Height
chr9	117552884	117552885	rs3810936	T	C	18	14	5.97E-01	8.50E-01	Crohns disease
chr9	117566439	117566440	rs4263839	A	G	65	80	2.45E-01	5.54E-01	Crohns disease
chr9	117568765	117568766	rs6478109	A	G	148	134	4.39E-01	7.66E-01	Inflammatory bowel disease
chr9	119134795	119134796	rs7869550	A	G	21	42	1.11E-02	6.26E-02	Height
chr9	121359285	121359286	rs11789399	G	A	4	14	NA	NA	Bipolar disorder and schizophrenia
chr9	123652897	123652898	rs881375	T	C	113	225	1.11E-09	4.99E-08	Rheumatoid arthritis
chr9	123690238	123690239	rs3761847	G	A	10	12	8.32E-01	1.00E+00	Rheumatoid arthritis
chr9	129465324	129465325	rs867559	A	G	5	7	NA	NA	BMI
chr9	130107963	130107964	rs4130590	A	G	4	7	NA	NA	Bipolar disorder
chr9	132370359	132370360	rs10988449	C	T	57	50	5.62E-01	8.44E-01	Response to antidepressants-bupropion
chr9	133464083	133464084	rs7466269	A	G	3	9	NA	NA	Height
chr9	136149398	136149399	rs507666	G	A	10	17	2.48E-01	5.54E-01	ICAM1
chr9	136925662	136925663	rs11789898	G	T	4	9	NA	NA	Platelet count
chrX	115302191	115302192	rs1403543	G	A	11	23	5.76E-02	2.16E-01	Cystic fibrosis severity

Table S10. GWAS SNPs at high-information positions in TF recognition sequences.

Transcription factor binding motifs overlapping GWAS SNPs were selected which had a large predicted difference in binding affinity between alleles, had a transcription factor consistent with the disease/trait class according to Gene Ontology annotation and which occurred in a DHS in a cell type compatible with the disease. Sequence logos derived from position weight matrices from the TRANSFAC and Jaspar databases. Box indicates position of GWAS SNP.

SNP	Target celltype	GO factor name	GWAS disease classes	GWAS diseases	Motif logo
chr9:117,568,766	GM06990;GM12864;GM12865; Jurkat;hTH1;hTH2	HOXA7	Autoimmune_disease	Inflammatory bowel disease	
chr5:150,458,146	GM12864;GM12865	ARNT	Autoimmune_disease	Systemic lupus erythematosus	
chr1:117,100,957	CD4;GM12864;GM12865; GM12878	IRF9	Autoimmune_disease	Multiple sclerosis	
chr8:11,352,541	GM12864;GM12865;GM12878	IRF8	Autoimmune_disease	Systemic lupus erythematosus	
chr5:110,435,490	CD56	ETV4	Hematological_parameters	Plasma eosinophil count	
chr4:77,420,784	GM06990;GM12864	AHR	Autoimmune_disease	Eosinophilic esophagitis (pediatric)	
chr4:27,744,718	fBrain	MEF2A	Neurological_behavioral	Brain imaging	
chr4:27,744,718	fBrain	ELK1	Neurological_behavioral	Brain imaging	
chr22:30,592,487	CACO2;GM06990	ATF1	Autoimmune_disease	Crohns disease	
chr6:135,426,573	K562	SIX1	Hematological_parameters	Mean corpuscular volume WBC count	
chr7:50,269,672	GM12864;GM12865	IRF3	Autoimmune_disease	Crohns disease	
chr8:11,352,541	GM12864;GM12865;GM12878	IRF9	Autoimmune_disease	Systemic lupus erythematosus	
chr20:8,114,704	BE_2_C;fBrain	VSX1	Neurological_behavioral	Colorword	
chr14:72,883,874	BE_2_C	EP300	Neurological_behavioral	Smoking_cestration	
chr11:131,320,070	hESCTO	MAX	Neurological_behavioral	Asperger disorder	
chr11:10,669,228	CD34;K562HL60	SMAD3	Hematological_parameters	Platelet aggregation-ADP	
chr19:34,168,273	BE_2_C;SKNSH	POU3F2	Neurological_behavioral	Working memory	
chr15:36,293,605	fBrain	REST	Neurological_behavioral	ADHD	
chr12:90,013,089	HBMEC;HCM	E2F4	Cardiovascular	Systolic blood pressure	
chr1:206,939,904	CACO2;CD14;CD34;CD4;CD56; CMK;GM06990;GM12864; GM12865;GM12878;hTH1;hTH2	EGR1	Autoimmune_disease Diabetes	Crohns disease Ulcerative colitis Type_1_diabetes	
chr3:39,555,580	fBrain	SOX14	Neurological_behavioral	ADHD	
chr11:131,320,070	hESCTO	GCM1	Neurological_behavioral	Asperger disorder	
chr14:69,408,697	hESCTO	GLI1	Neurological_behavioral	Smoking behavior	
chr1:117,100,957	CD4;GM12864;GM12865; GM12878	IRF9	Autoimmune_disease	Multiple sclerosis	
chr4:15,397,906	fBrain	PBX1	Neurological_behavioral	Conduct disorder (case_status)	
chr8:34,126,948	fBrain	FOXA1	kidney_lung_liver_neural	Neurological behavioral	

Table S10. GWAS SNPs at high-information positions in transcription factor motifs. (cont'd)

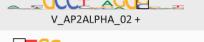
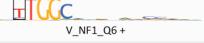
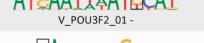
SNP	Target celltype	GO factor name	GWAS disease classes	GWAS diseases	Motif logo
chr4:15,397,906	fBrain	PBX1	Neurological_behavioral	Conduct disorder (case_status)	
chr3:60,771,108	BE_2_C	TFAP2A	Neurological_behavioral	ADHD	
chr3:32,842,101	hESCTO	NR2F2	Neurological_behavioral	ADHD	
chr6:139,839,423	CMK:fHeart	EGR1	Hematological_parameters	Mean corpuscular volume	
chr2:12,568,996	HAsp	FOXA2	Neurological_behavioral	Bipolar disorder and schizophrenia	
chr21:17,828,291	HAsp	PPARG	Neurological_behavioral	ADHD	
chr15:68,198,911	BE_2_C;HAc	STAT3	Neurological_behavioral	Trails B	
chr2:11,220,562	fBrain	TFAP2A	Neurological_behavioral	PC1	
chr1:11,852,516	fHeart	NF1	Cardiovascular	Atrial fibrillation	
chr1:76,484,014	BE_2_C	POU3F3	Neurological_behavioral	Novelty seeking	
chr17:38,051,348	CD56;hTH2	SPI1	Autoimmune_disease	Ulcerative colitis	
chrX:44,400,186	fBrain	POU3F2	Neurological_behavioral	Personality dimensions	
chr7:50,269,672	GM12864;GM12865	GATA4	Autoimmune_disease	Crohns disease	
chr6:142,750,516	SAEC	RORA	Kidney_lung_liver	FEV1/FVC	
chr11:131,320,070	hESCTO	GCM1	Neurological_behavioral	Asperger disorder	
chr16:20,367,690	fKidney	SP1	Kidney_lung_liver Serum_metabolite	Chronic kidney disease Creatinine	
chr1:93,401,837	fBrain	PAX6	Neurological_behavioral	Bipolar disorder and schizophrenia	
chr17:72,268,208	fBrain	KLF7	Neurological_behavioral	Information processing speed (inspection_time)	

Table S11. Cell-selective enrichment of Crohn's disease-associated variants.

Fold-enrichment and significance levels (*P*-value, Fisher's exact test) for DHSs of each tested cell type for GWAS SNPs associated with multiple sclerosis at varying thresholds (association $P < 10^{-3}$, 10^{-10} , and 10^{-20}).

Cell_type	Enrichment (GWAS $P < 10^{-3}$)	Fisher_P- value	Enrichment (GWAS $P < 10^{-10}$)	Fisher_P- value	Enrichment $P < 10^{-20}$	(GWAS Fisher_P- value)
Th17	2.73	3.74E-45	4.53	7.37E-11	11.95	5.39E-13
Th1	2.6	1.06E-43	4.73	1.06E-12	8.87	3.30E-09
CD56+	2.48	6.80E-82	4.06	2.17E-19	6.03	3.73E-10
CD8+	2.83	7.47E-74	3.92	8.38E-12	6.02	8.42E-07
Th2	2.36	1.35E-53	3.73	1.29E-12	5.88	1.29E-07
CD4+	2.71	4.44E-83	4.23	6.24E-17	5.58	2.43E-07
CD3+	2.77	5.09E-67	4.2	5.54E-13	5.24	2.37E-05
CD3+_CordBlood	2.76	4.44E-59	4.45	4.94E-13	4.69	0.000334049
CD14+	2.42	7.17E-74	3.98	4.40E-18	4.27	1.21E-05
CD34+	2.03	5.39E-37	3.66	1.91E-13	4.13	7.94E-05
GM06990	2.38	2.13E-47	3.05	1.87E-07	3.89	0.0011313
CD19+/CD20+	2.65	1.20E-84	4.79	3.87E-23	3.68	0.000416989
GM12865	2.19	1.23E-54	3.11	6.48E-11	3.59	0.000263786
GM12864	2.27	2.09E-52	3.05	3.81E-09	3.42	0.001344248
GM12878	2.75	6.58E-84	3.38	1.46E-10	3.28	0.003220858
fSpleen	1.94	3.43E-52	3.26	7.92E-17	3.19	0.000129487
HMVEC_dLyAd	1.59	1.65E-14	1.96	0.00160193	3.02	0.005248111
vHMEC	1.52	8.28E-17	2.08	2.46E-05	2.52	0.006518382
HMVEC_LLy	1.6	9.25E-18	2.23	2.13E-05	2.5	0.01530051
fIntestine_Lg	1.49	3.16E-45	2.16	2.58E-15	2.36	1.74E-05
HMEC	1.59	5.92E-16	2.32	1.33E-05	2.35	0.03059794
HEEPiC	1.53	2.66E-18	1.85	0.000458429	2.19	0.02219043
fIntestine_Sm	1.57	2.10E-51	2.26	2.53E-15	2.18	0.000374271
fThymus	1.68	8.10E-62	2.44	7.73E-17	2.11	0.001449633
HCPEpiC	1.48	2.81E-16	1.51	0.01865873	2.08	0.02942868
HMVEC_dAd	1.63	1.08E-14	2.15	0.000473185	2.07	0.09572298
NHEK	1.8	1.42E-26	1.55	0.03350289	2.05	0.07427661
HPAEC	1.51	9.38E-11	2.12	0.000574955	2.04	0.1000967
HIPEpiC	1.5	4.62E-18	1.94	6.46E-05	2.02	0.03516575
HMVEC_dNeo	1.59	3.19E-16	2.08	0.000225119	1.98	0.08355248
SAEC	1.46	9.95E-14	1.57	0.01350666	1.82	0.0922865
HMVEC_dLyNeo	1.48	7.29E-13	2.07	0.000110797	1.8	0.1190631
HMVEC_LBl	1.53	9.84E-18	2.28	1.00E-06	1.75	0.1067207
SkMC	1.37	4.61E-10	1.64	0.00560093	1.71	0.1170378
IMR90	1.49	2.00E-16	1.51	0.02029439	1.68	0.1249528
HSMM	1.34	2.07E-11	1.54	0.006564184	1.68	0.08908063
HNPCePiC	1.44	5.67E-14	1.47	0.02662905	1.64	0.1365095
HRE	1.46	4.56E-13	1.58	0.01434599	1.62	0.1666523
HMVEC_dBlAd	1.57	5.97E-19	2.3	1.47E-06	1.59	0.1774125
PrEC	1.48	5.06E-12	1.38	0.09546684	1.59	0.2068589
HBMEC	1.39	1.20E-13	1.86	7.79E-05	1.56	0.1426867
HUVEC	1.42	6.35E-10	2.22	2.48E-05	1.55	0.2219272
HRPEpiC	1.27	6.02E-07	1.6	0.005558879	1.52	0.1791802
fPlacenta	1.65	8.00E-47	1.99	6.50E-08	1.48	0.1225238
fLung	1.28	9.01E-21	1.46	9.04E-05	1.46	0.04820113
HFF_MyC	1.46	1.15E-16	1.63	0.003290356	1.46	0.2040778
HCFaa	1.48	2.76E-18	1.69	0.001306412	1.44	0.2141146
WI_38	1.55	1.24E-16	1.55	0.02157066	1.43	0.2700086
fKidney	1.23	2.21E-18	1.52	8.47E-07	1.36	0.06992943
HRGEC	1.58	9.68E-16	2.11	0.000168924	1.35	0.3464
HMVEC_dBlNeo	1.58	7.97E-23	2.12	3.20E-06	1.34	0.2930424
HAsp	1.28	5.99E-09	1.25	0.1035101	1.32	0.2597848
fStomach	1.5	2.52E-37	1.85	6.03E-08	1.27	0.2371028
AG04449	1.42	6.22E-10	1.2	0.2432996	1.24	0.4065483
NHDF_Ad	1.39	6.03E-13	1.34	0.06100607	1.2	0.3833231
fAdrenal	1.34	4.37E-15	1.46	0.005624388	1.1	0.4437991

Cell_type	Enrichment (GWAS P<10 ⁻³)	Fisher_P- value	Enrichment (GWAS P<10 ⁻¹⁰)	Fisher_P- value	Enrichment (GWAS P<10 ⁻²⁰)	Fisher_P- value
Trophoblast	1.33	3.50E-06	1.46	0.06784831	1.06	0.5380353
HAc	1.35	5.32E-09	1.17	0.2646637	1.03	0.5492765
AG09309	1.37	1.08E-09	1.38	0.06990271	1.02	0.5572025
HSMM_D	1.4	8.61E-17	1.87	1.10E-05	0.9792	0.579658
HPAF	1.47	7.23E-15	1.26	0.1495196	0.9688	0.59537
WI_38_TAM	1.45	3.24E-17	1.51	0.01014413	0.9683	0.5925241
HCM	1.53	1.40E-18	1.39	0.05509522	0.952	0.6092184
fMuscle	1.29	1.61E-30	1.47	2.49E-06	0.9472	0.6388238
fSkin	1.25	1.62E-11	1.39	0.006067741	0.9452	0.6193618
NHLF	1.32	7.08E-09	1.32	0.08840022	0.8669	0.6822042
HCF	1.51	3.17E-15	1.27	0.1613846	0.8361	0.6998518
hESCT0	1.23	1.12E-05	1.26	0.1287957	0.8296	0.715168
fSpinal_cord	1.06	0.0508844	1.1	0.261125	0.8119	0.7789466
BE_2_C	1.19	0.000800551	0.9482	0.6216681	0.7722	0.7498395
HPdLF	1.46	1.11E-13	1.55	0.0157039	0.7684	0.752791
HRCE	1.44	5.78E-13	1.27	0.1450191	0.7658	0.7548389
AoAF	1.48	8.41E-15	1.26	0.1562488	0.7577	0.7612351
HVMF	1.56	3.78E-19	1.58	0.01153608	0.7543	0.7639403
HAEpiC	1.51	5.90E-17	1.66	0.004877783	0.7395	0.7756365
RPTEC	1.43	1.80E-12	1.44	0.03903286	0.7394	0.7757033
AG04450	1.56	6.87E-14	1.39	0.1059331	0.7106	0.77538
HGF	1.43	2.40E-09	1.19	0.2752837	0.6862	0.7917667
iPS_19_11	1.42	1.80E-09	1.52	0.03983261	0.6694	0.8031659
HFF	1.51	5.60E-19	1.53	0.01302448	0.6603	0.8371519
HPF	1.59	3.32E-17	1.61	0.01667762	0.6193	0.8368622
iPS_4_7	1.33	3.80E-07	1.07	0.4157521	0.5834	0.8605886
HMF	1.43	4.06E-11	1.24	0.1891547	0.5743	0.8665022
fHeart	1.26	2.76E-16	1.27	0.01889588	0.5637	0.9711566
HAh	1.39	7.24E-14	1.32	0.06613878	0.5539	0.9118636
Mesendoderm	1.27	2.88E-06	1.35	0.08213618	0.4988	0.9131563
NHA	1.32	2.18E-08	1.31	0.1039998	0.4633	0.9328191
fBrain	0.9483	0.956259	0.7322	0.9910235	0.3839	0.9975966
HESC	1.47	6.12E-10	1.48	0.06778897	0.3787	0.930943
AG09319	1.4	3.14E-08	1.1	0.3975671	0.3623	0.9389211
HConF	1.59	8.01E-16	1.26	0.2029553	0.3407	0.9489365
iPS_6_9	1.36	3.81E-08	1.32	0.1323166	0.3037	0.9646771
BJ	1.39	3.16E-10	1.15	0.3001724	0.2642	0.9787703
NHDF_Neo	1.24	3.29E-05	0.9054	0.6945273	0.2457	0.9841808
fTestes	1.37	5.81E-11	1.3	0.1012356	0.2225	0.9898362
NPC	1.63	1.52E-13	1.99	0.003289018	0	1
H1_P18	1.41	1.41E-10	1.42	0.0630504	0	1
H9_P42	1.46	2.10E-10	1.46	0.06742848	0	1
AG10803	1.35	9.20E-08	1.02	0.5020398	0	1
iPS_19_7	1.32	6.66E-06	0.9502	0.610338	0	1

Table S12. Cell-selective enrichment of multiple sclerosis disease-associated variants.

Fold-enrichment and significance levels (*P*-value, Fisher's exact test) for DHSs of each tested cell type for GWAS SNPs associated with multiple sclerosis at varying thresholds (association *P* < 10⁻³, 10⁻¹⁰, and 10⁻⁷⁵).

Cell_type	Enrichment (GWAS <i>P</i> <10 ⁻³)	Fisher_ <i>P</i> - value	Enrichment (GWAS <i>P</i> <10 ⁻¹⁰)	Fisher_ <i>P</i> - value	Enrichment (GWAS <i>P</i> <10 ⁻⁷⁵)	Fisher_ <i>P</i> - value
CD3+_cb)	4.78	7.58E-222	8.4	2.95E-119	10.23	3.35E-17
CD19+/CD20+	4.26	1.53E-279	6.72	1.27E-129	7.79	1.67E-17
GM12878	3.87	1.79E-202	5.75	1.01E-86	6.31	3.12E-11
CD14+	3.08	2.48E-152	4.35	5.66E-64	5.44	6.10E-11
GM06990	3.52	8.06E-143	4.36	2.65E-44	5.16	3.07E-07
GM12865	3.13	9.98E-159	3.63	1.65E-43	3.78	6.59E-06
GM12864	3.38	6.53E-163	3.92	4.11E-44	3.31	0.000326316
CD56+	3.41	4.66E-193	3.85	2.40E-49	3.09	0.000350238
CD8+	4.21	4.79E-197	5.51	8.66E-65	2.96	0.006159673
CD4	3.81	3.82E-198	4.63	1.50E-57	2.68	0.007038724
Th1	3.58	1.41E-101	4.05	2.56E-26	2.51	0.05062207
fSpleen	2.28	1.40E-94	2.7	1.79E-30	2.5	0.001122999
Th2	3.13	7.64E-122	3.12	3.79E-24	2.44	0.01809435
CD34+	2.47	1.03E-74	3.03	1.14E-25	2.4	0.01381383
NPC	2.01	1.08E-32	2.34	3.45E-11	1.95	0.09072312
HPAEC	1.9	3.93E-29	2.35	2.21E-12	1.77	0.1252959
HMF	1.46	1.74E-13	1.43	0.001866852	1.66	0.113286
Th17	3.41	2.83E-83	3.72	1.65E-20	1.64	0.2777823
fTestes	1.5	3.16E-19	1.61	1.64E-06	1.59	0.1007764
CD3+	3.94	2.67E-161	4.74	5.57E-46	1.56	0.2546488
H1_P18	1.77	1.99E-33	2.26	3.18E-16	1.55	0.1482745
HCM	1.67	4.49E-30	1.71	1.62E-07	1.55	0.1315509
AG10803	1.38	1.67E-09	1.38	0.006081542	1.5	0.1894908
H9_P42	2.09	1.47E-48	3.14	3.72E-30	1.47	0.2256847
HMVEC_dB1Neo	1.83	1.74E-45	2.12	6.33E-16	1.46	0.1677198
HCF	1.55	4.57E-18	1.53	0.000176767	1.41	0.228986
HPAF	1.57	2.31E-22	1.57	1.65E-05	1.4	0.2159259
HMVEC_dLyAd	1.87	4.06E-30	2.12	5.94E-10	1.36	0.3067913
HMVEC_LLy	1.8	4.80E-31	1.92	9.40E-09	1.35	0.2874307
HUVEC	1.74	1.04E-27	1.73	2.24E-06	1.34	0.2939138
HPF	1.59	1.72E-18	1.27	0.0403532	1.34	0.2933387
HMVEC_dB1Ad	1.83	2.61E-39	1.95	1.17E-10	1.33	0.273031
AG09319	1.52	5.79E-13	1.35	0.01921505	1.32	0.3306616
HPdLF	1.39	3.44E-11	1.32	0.01074998	1.3	0.2940502
HFF	1.54	3.64E-22	1.53	2.89E-05	1.29	0.2807983
vHMEC	1.77	1.29E-35	1.98	1.43E-11	1.28	0.3055248
AoAF	1.38	7.86E-11	1.21	0.05987165	1.28	0.3069095
HESC	1.86	2.10E-31	2.1	1.81E-10	1.26	0.3637511
iPS_6_9	1.88	8.12E-40	2.37	1.56E-17	1.24	0.3545499
HEEPiC	1.66	1.67E-28	1.63	3.77E-06	1.24	0.3374921
iPS_19_7	1.73	1.59E-24	1.94	2.10E-08	1.23	0.3818447
HFF_MyC	1.55	4.91E-24	1.43	0.000367984	1.22	0.3370015
HMEC	1.89	1.84E-35	1.86	1.45E-07	1.2	0.4065026
fThymus	1.9	1.36E-112	1.82	3.18E-19	1.18	0.2911422
HCfaa	1.45	2.00E-17	1.3	0.007857596	1.18	0.370445
fIntestine_Sm	1.77	2.28E-95	1.77	4.51E-19	1.14	0.3313635
iPS_19_11	1.84	5.19E-34	2.42	4.88E-17	1.13	0.4548328
HIPEpiC	1.48	9.92E-18	1.2	0.06168806	1.13	0.4210396
HRCE	1.55	4.56E-20	1.61	8.49E-06	1.11	0.4545429
AG09309	1.34	2.87E-09	1.27	0.02527731	1.1	0.4632312
fAdrenal	1.49	1.32E-29	1.42	1.68E-05	1.09	0.4305533
RPTEC	1.53	3.11E-19	1.59	1.52E-05	1.08	0.4825215
HAEpiC	1.5	8.50E-18	1.38	0.002803663	1.06	0.4961963
NHDF_Neo	1.31	3.48E-08	1.08	0.2717603	1.06	0.5039732
WI_38	1.52	3.06E-16	1.36	0.00793145	1.04	0.5286493

Cell_type	Enrichment (GWAS P<10 ⁻³)	Fisher_P- value	Enrichment (GWAS P<10 ⁻¹⁰)	Fisher_P- value	Enrichment (GWAS P<10 ⁻⁷⁵)	Fisher_P- value
AG04450	1.67	3.50E-20	1.39	0.009287482	1.03	0.5442849
HCPePiC	1.54	6.33E-21	1.32	0.007900789	1.01	0.5515751
HNPCePiC	1.39	4.01E-12	1.1	0.2184781	1.01	0.544658
Trophoblast	1.46	4.89E-11	1.61	0.000154718	1.01	0.5586931
iPS_4_7	1.61	7.52E-22	1.98	9.16E-11	0.9954	0.5661271
fMuscle	1.19	3.46E-16	1.03	0.3260101	0.9909	0.5525155
HConF	1.67	7.11E-21	1.76	3.55E-06	0.9894	0.5771841
fKidney	1.26	1.23E-24	1.2	0.000320705	0.9892	0.5561045
fStomach	1.58	3.45E-52	1.35	2.95E-05	0.9875	0.5597261
HRGEC	1.66	7.58E-21	1.74	5.09E-06	0.979	0.5855181
BJ	1.38	1.96E-10	1.23	0.05234849	0.953	0.6047794
HAh	1.36	7.39E-13	1.22	0.02966251	0.938	0.621058
fIntestine_Lg	1.7	6.04E-91	1.58	8.68E-14	0.9346	0.6462106
HAc	1.4	1.07E-11	0.9487	0.6713378	0.9271	0.6290546
HVMF	1.45	2.74E-14	1.16	0.1216169	0.9141	0.6413908
HMVEC_LBl	1.84	1.57E-41	1.79	2.11E-08	0.9075	0.6476765
AG04449	1.45	4.20E-12	1.25	0.04979334	0.8964	0.6544684
HMVEC_dAd	1.86	1.07E-26	1.72	4.73E-05	0.8951	0.6533748
SkMC	1.27	1.30E-06	1.06	0.3332893	0.8745	0.6794557
NHDF_Ad	1.23	2.78E-06	1.24	0.02451941	0.8717	0.6890466
IMR90	1.49	2.01E-17	1.25	0.03304745	0.8646	0.6891373
Mesendoderm	1.57	1.20E-22	1.69	3.83E-07	0.8638	0.6898794
WL_38_TAM	1.49	6.04E-21	1.22	0.03139724	0.8509	0.7110976
HBMEC	1.34	2.40E-11	1.07	0.2936797	0.8334	0.7295428
fSkin	1.17	6.94E-07	0.9938	0.5431454	0.8182	0.794983
HSMM	1.22	4.06E-06	1.03	0.4114304	0.8046	0.7597594
fSpinal_cord	1.05	0.08495058	0.7891	0.997174	0.8037	0.813415
fPlacenta	1.45	5.14E-26	1.22	0.01323427	0.7715	0.8193974
SAEC	1.64	2.52E-25	1.46	0.000634212	0.7498	0.7832075
HGF	1.49	2.96E-12	1.39	0.008078605	0.7482	0.766667
fLung	1.27	2.35E-22	1.06	0.1837693	0.7228	0.9335765
HSMM_D	1.34	1.65E-13	1.13	0.1120233	0.7022	0.8601957
fBrain	0.9138	0.9986694	0.6629	1	0.6458	0.965557
fHeart	1.18	4.87E-09	0.79	0.9991676	0.6282	0.9683334
HRPEpiC	1.28	7.28E-08	0.9487	0.68418	0.6162	0.8920638
HAsp	1.2	9.37E-06	0.7857	0.9863805	0.584	0.933101
BE_2_C	1.34	4.50E-09	1.16	0.1272647	0.5379	0.9196424
NHEK	2.08	3.53E-47	2.17	1.30E-11	0.4989	0.9115511
NHA	1.28	2.02E-07	0.9268	0.7411457	0.4973	0.9424616
PrEC	1.62	1.40E-19	1.37	0.009063974	0.4589	0.9337129
HMVEC_dLyNeo	1.76	7.83E-30	1.68	6.95E-06	0.4318	0.9473133
hESCT0	1.32	2.04E-10	1.21	0.04395004	0.4287	0.9725312
HRE	1.63	2.06E-23	1.43	0.001611954	0.3906	0.9652407
NHLF	1.25	1.07E-06	1	0.5046302	0.3115	0.9889289
HMVEC_dNeo	1.84	2.85E-32	1.88	7.64E-08	0.2391	0.9854898

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